TEACHING UNITS ON GENETICS:
MOTIVATION AND THE APPLICATION OF KNOWLEDGE

By

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ABSTRACT

There are three steps in planned learning: motivation, activity, and reinforcement. This project details the problems encountered by teachers when they attempt to motivate and reinforce student learning. These problems in turn result in an emphasis on the selection of an activity.

Of all the factors in motivation, it is proposed that an increase in one of them--the willingness to learn--will have a greater effect on learning than the choice of an activity, and that this increase can be accomplished by showing the students how the content to be learned can be immediately useful to them.

The genetics unit from the grade thirteen biology course is amended in order to illustrate just how a teacher can alter a course to make it more useful to the students and yet still meet any future academic requirements. By having students apply theoretical knowledge to resolve life problems, it is suggested that student willingness to study is enhanced and the overall motivation to learn is significantly increased.

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CHAPTER ONE

INTRODUCTION

It is often said in one form or another that people learn better when they are interested in what they are studying. Certainly everyone in education has seen evidence that this is at least frequently true, and no doubt most, if not everyone, whether in education or not, has experienced or seen faster and/or more thorough learning take place when the learner wanted to develop a skill or acquire some body of knowledge.

I feel that this generally accepted fact is unfortunately de-emphasized by both teachers and curriculum designers, the persons who should be making the most of its existence.

At times, interest in a topic is not enough to increase learning for there are other factors involved. (I shall discuss these other factors later.) But two relationships which highlight the importance of interest to learning can be identified.

As the desire to learn increases, the importance of most other factors decreases; and as the usefulness (i.e. relevance of the knowledge) increases so does the desire to learn it.

In the United States, the Biological Sciences Curriculum Study Committee has stated, "Education is more than just the transmission of facts. It involves the presentation of information in a context that has relevance for the prospective learner."[1] It is the recognition of

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[1] BSCS Advisory Committee, "A Prepared Statement for the Public Health Service Genetics Coordinating Committee", The Biological Sciences Curriculum Study Journal, (I, February, 1978), p. 31.

relevance that students need.

The desire to learn is called willingness by Sorenson[2] and he considers it to be only one of five motivating factors. As a factor in student motivation it should be one of the aims of all teachers to increase its level as much as possible whether they feel that it is the main factor in learning or not.

The second of Sorenson's factors of motivation is readiness, whereby the students require certain capacities or abilities. These abilities must be matched to the learning task at hand. No one can seriously expect a grade nine student to get much understanding from a university text on quantum mechanics no matter how much he wants to learn it, but on the other hand for motivation the learning task cannot be so easy as to be boring and unsatisfying to the learner.

The third factor is the aspiration level, and it refers to both the difficulty of the learning tasks a person is willing to undertake, and the amount of work he is willing to try to do on them in a given time. Small successes gradually result in a rise in this aspiration level and a greater desire to keep succeeding.

The fourth factor is that of attitude. Basically it means that if a student likes the situation he is in, he will do what is necessary to maintain that situation.

Sorenson's fifth and final factor is anxiety. He feels this is a negative influence for it causes a dislike for the situation in which the student finds himself, and distracts the student from concentrating

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[2]H. Sorenson, Psychology in Education (McGraw-Hill, 1964), p. 407.

on the tasks at hand. The only time it has a positive effect is in the performance of simple mechanical tasks.

Anxiety should not be confused with tension, which Sorenson feels is due to the excitement of working towards completion of a job, or the feeling that one is learning something that they want to know. Tension will disappear when a task is finished, and its reduction is a form of internal reinforcement.

One may not agree that these are all the factors involved in motivation and because of the overlap and effect they have on each other, it is possible to re-organize or to subdivide them as some other writers have done. However, most of this re-organization occurs in name only; the same phenomena are involved.

Unfortunately, a teacher who tries to use all of the above motivating factors will be unable to emphasize any one of them. Because willingness does not appear to be easy to promote in a classroom, it will frequently be de-emphasized, neglected, or more commonly be replaced with a more "interesting" activity. (More will be said about such activity in Chapter Two.)

For student learning, the other means of motivation are also difficult to promote so that attempts to employ these may not result in the behaviour one desires, i.e. specific learning.

I shall attempt to explain in this chapter how willingness (to learn) can be increased by emphasizing the practicality, or useful nature, of the material under study, and shall leave any discussion of problems with the other motivational means until chapter two.

Throughout the remainder of this project I shall reserve the words "relevant" and "relevance" when used by myself to mean likely to

be useful, in the opinion of the students, for the students' behaviour and decisions both outside and within the school. It is the viewpoints of the students that must be considered, and not only those of the teachers, when motivation is the concern.

No doubt there are many teachers and course designers who think that they have, or are, presenting material that is useful when in fact it is not. These "uses" are such from a teacher viewpoint and can be grouped into the following categories: that which provides a general background knowledge (i.e. a scientific literacy); that which encourages a student to apply facts and behaviours to solve problems; that which qualifies a student for admission to a job or advanced level of education; that which the student can actually use in the performance of a future vocation or in the study of future academic material; and that which the student can use in his or her life outside of school.

Any and all of these can be very real uses depending on a particular student's situation, but unfortunately the emphasis has been on obtaining general knowledge and prerequisite knowledge for future academic study, ignoring Dewey's work.

Years ago, John Dewey recognized the problem of having curriculum designed and taught by specialists, for the roles of a scientist and a science teacher are quite different. As he puts it, "His (the teacher's) problem is that of inducing a vital and personal experiencing. Hence, what concerns him, as teacher, is the ways in which that subject may become a part of experience; what there is in the child's present that is useable with reference to it."[3]

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[3] J. Dewey, "The Logical and Psychological Aspects of Experience. "Theory of Knowledge and Problems of Education, ed. D. Vandenberg (Chicago: U. Illinois Press, 1969), pp. 187-188.

This quotation emphasizes a second point about the usefulness of material--it must be useable in the "present".

This is why I do not consider the acquisition of a general background of knowledge a use. Out of all the pieces of information gathered through years of schooling very few are retained long enough or in detail enough to actually be used should the opportunity eventually arise. One cannot expect students to be motivated to learn material once they realize this for themselves.

Neither have I included the attainment of some arbitrary grade to quality for a job, etc. as a use, for this will only be true in the case of a senior student about to apply (in the very near future) for a job or for post-secondary school education.

Experience has shown that telling a student who is far from graduating that he will need to learn something for future use does not provide any incentive to study, and if a student is doing poorly and needs more motivation, stating job or education requirements leads to depression and frustration more often than it leads to motivation.

Knowledge that can be applied to the performance of a job is beneficial, but is greatly exaggerated for very little of the total knowledge acquired throughout secondary school fits this category.

In their article "Education and the Uses of Knowledge", Broudy, Smith and Burnett argue against the notion that schooling is job training, for in their view, the knowledge obtained will not be used. The authors observe that, "We hear the argument that certain subjects should be taught in high school because they will be used in certain vocations. .....We are often ruefully surprised at how little of our schooling we

actually use on the job."[4] They hold that schooling gives one a general background from which one may draw generalizations but only on occasion specific items.

It is interesting to note that in contrast to their view, one of the objectives of the Secondary Education Review Project is "to redesign the (secondary school) program to better prepare students for the world of work."[5]

Of course knowledge needed for the performance of a job, or for future education may supply motivation in the form of increased student willingness. But this will be true only if the student <u>believes</u> the knowledge is soon to be needed. Thus the appeal to a "future" use which is employed by many teachers will work on occasion and is more likely to be successful in the higher grades and universities. Here the "future" usually alluded to is after school life and it is less remote from the present.

Any course or programme must have a goal and I feel that providing students with prerequisite knowledge to go further and further in the study of science is the main goal of science courses. This may sound contradictory to my stated views on the usefulness of material but the goals are not mutually exclusive. In reaching this goal of preparing for future studies all of the "uses" previously mentioned will be met. But for motivational purposes, and in recognition of the fact that the majority of the students will not make a career of science, it is

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[4] H. Broudy, B.O. Smith, J.R. Burnett, "Education and the Uses of Knowledge". <u>Theory of Knowledge and Problems of Education</u>, ed. D. Vandenberg (Chicago: U. of Illinois Press, 1969), p. 68-69.

[5] Secondary Education Review Project description pamphlet by Ministry of Education, (1981), p. 4.

essential that as much factual, conceptual or behavioural content as possible be related to the students' present lives and experience outside of school.

As it stands now, subject matter is usually presented as a given body of truth, the way a scientist sees it, without regard for the pupils' present experience. Thus the students may find themselves in situations where they understand the individual words but do not grasp well enough the sentences or thoughts formed by the words so as to see how to apply them to themselves or their worlds.

Relating material to students' experiences so they can apply it, may sound like a very minor change, and in a few units it is, but alterations can be quite extensive when one considers entire courses.

Agreeing with the statement of Dewey given earlier, Newbury[6] also states that if the subject matter is removed from the learner and his needs, it cannot sustain the learner's interest and attention. Nevertheless, she criticizes "progressive educators" who give the child something to do which interests him without regard to the subject matter.

This is not my intention. One should not alter the content merely to please the learner, nor should the student choose the content to any great extent.

Since more and more material has been transferred to the lower grades, it is time to consider a complete re-allocation of the content material. Just because a topic has always been taught in grade ten does not mean that it should stay there or move to grade nine. Maybe it

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[6] D.J. Newbury, "The Discipline in Dewey's Theory of Inquiry." Theory of Knowledge and Problems of Education, ed. D. Vandenberg (Chicago: U. of Illinois Press, 1969), p. 264.

should be delayed until grade twelve or thirteen or in some cases dropped altogether. Unfortunately, if a topic is easy enough to be understood in the lower grades there appears to be an aversion to reserving it until the senior grades or even college.

Once the content has been established, assuming it has been chosen on some reasonable basis, then the effort must be made to make its immediate usefulness apparent to the students.

Because of individual differences the applications of any particular topic are not going to appeal to everyone but much of the content can be made more interesting to the students by relating it to interests that they already have outside the classroom.

One example of this is a course established by W.S. Franz at Walter Johnson High School in Bethesda Md. where students learn "applied science"[7]. He attributes part of his success at changing behaviour, attendance and learning, to the practical subject matter dealt with and part to the methodology used at the same time. The subject matter is not just practical however; for it can be applied immediately as well as in the future.

As for the difficulty in finding uses that are recognized as such by the students, one partial solution stems from the idea of "following through" with a topic. Every item of information or skill along the way does not need to be of personal interest, but the end result of the unit must leave the students with the feeling that they have gained something for themselves. If this feeling is ever reached in academic courses it

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[7] W.S. Franz, "Applied Science Moves the Unmotivated", The Science Teacher, XLVI (6), (September 1979), p. 36-38.

is at the end of the year or at graduation which is far too long to serve as a goal for most students.

As an example, consider the "chemistry" taken presently in grade nine. According to the curriculum guideline[8], the students are to learn material such as definitions of atoms, elements, molecules, ions, to name and give symbols of a few specified elements, and to begin to see element similarities from a periodic table. A little of this material is used in the course to explain some behaviours of materials, but the majority of it is non-useable by the students, for the topic stops and study of a new one starts. Thus they cannot use what they have learned academically, nor can they apply it to their lives for it is far too shallow.

According to this guideline, "The focus of this unit is on the development of the intellectual skills of scientific inquiry"[9]. This is an admirable goal, but surely it could be met by using a topic of more use and therefore more interest to the students. How can one truly expect the students to be motivated to develop their skills, when the answers (i.e. facts) that they obtain by using them are useless by themselves, and do not appear to lead anywhere?

This is the type of unit that should be broken up, and presented only when each particular piece of information is going to be used to learn something else or carried far enough to include something that the student views as relevant.

As an example of a method to increase student willingness I have

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[8]Ministry of Education, Intermediate Division Science (1978), pp. 165-166.

[9]Ibid., p. 165.

chosen the unit on genetics from the grade 13 biology course, for as it is presently presented it is very similar to the unit of chemistry just described for the grade nine course. The content is given but there is no attempt made to make it relevant to the students.

The present directive[10] from the Ministry of Education treats genetics as a part of reproduction by dealing with Mendelian relationships, and then introducing the idea of mutations briefly and seemingly only to support and explain the theory of evolution.

Both the intent and content given are vague enough to allow a teacher some freedom of choice in each, but it seems that the genetics itself is merely introduced to explain diversity and only for purposes of future academic study. No effort is made to show that the topic has applications which should be of interest to present and future mankind.

A scientist might view this organization as an example of how evidence can be accumulated to create and support a theory, and hence he may consider this arrangement useful. But for the purpose of motivation such a treatment is inadequate.

With current issues of carcinogens, cloning, recombinant DNA, and birth abnormalities, the application of genetics to the understanding of these topics is relevant not only to the students' lives in the near future, but to the present as well. More important, this is true whether the students continue their education beyond high school or do not.

As will be shown in detail in Chapters Three and Four, while using the genetics unit as an example, I have kept the content and intent

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[10] Ontario Department of Education, Biology: Grade 13 (1969), pp. 13-14, 31.

prescribed by the Ministry, have made changes in the order of items and the emphasis, but have added more material to make the unit not only appear useful to mankind, but personally useful for the students in their lives outside of school.

This approach of showing usefulness is consistent with the BSCS committee findings. As they put it, "Information that has important personal implications for the learner is inherently more interesting and learned more thoroughly than isolated facts that have no practical application."[11]

In a letter to the Secondary Education Review Project from STAO, the following statements are made: "Many of the American science courses we have adopted in Ontario tend to emphasize the structure of the discipline, in depth learning of the subject content and considerable mathematical ability. To students who are looking for fulfillment of immediate objectives, job related learnings, practical applications of science, and relevance to everyday life these more academic courses have little appeal.... The students need to be able to use these facts in dealing with issues in everyday life."[12]

The letter goes on to say that, "Often the justification given for including the inappropriate material is that it will be needed later. For most students, "later" never comes."[13]

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[11] BSCS Advisory Committee, "A Prepared Statement for the Public Health Service Genetics Coordinating Committee", The Biological Sciences Curriculum Study Journal, (I, February, 1978), p. 32.

[12] Science Teachers' Association of Ontario, "Recommendations to the Science Education Review Project", The Crucible, XII, No. 2 (March, 1981), p. 5.

[13] Ibid., p. 6.

I believe that statements like these show that more and more science teachers are beginning to think, if not as strongly, in the same direction as I.

Methods of reinforcement and all the motivational factors have problems when implemented to increase learning. Besides those given in this chapter, other suggestions are made in Chapter Six concerning methods of dealing with the problems of increasing the willingness factor.

The next chapter will detail some of the difficulties that arise when attempts to use the other motivational factors and reinforcement are made. It is necessary to consider these before moving to the example for otherwise, the reader may do just what many teachers have done, and that is to think that the problems with these others are more easily overcome than those with willingness. To think this results in a neglect of willingness promotion and the eventual neglect of all serious motivation attempts when the problems with these other motivational factors are not easily overcome.

CHAPTER TWO

CONDITIONS OF LEARNING IN THE CLASSROOM

Given some content to be learned, whether it be factual or operational, there are three basic steps in the learning process: motivation of the students, activity by the students, and reinforcement of student behaviour.

Learning does occur in schools so these three steps must occur, but there are also students who learn very little and others who do not learn as much as they are able. There are also those whose behaviour causes problems for themselves and others, and those who leave school as soon as they are legally permitted. It seems then, that one or more of the steps in this simple list is not getting adequate attention.

Any theory of learning such as this list of steps represents is bound to have problems when one tries to apply it in a classroom, for this simple list fails to show the relationship between the components, and the intricate nature of each individual step.

It is my belief that due to problems that arise when trying to motivate and reinforce student behaviour, almost all of the emphasis is placed on the activity portion of this process.

The purpose of this chapter is to outline what these problems are. Many, if not all, of these will seem obvious when put into print, but it is necessary that they be recognized so that a comparison can be made between those of increasing willingness and those of the other motivational factors and reinforcement.

Before proceeding it is best to clarify the meaning of "activity"

and "content" as I am using them.

"Content" is the factual material or operation that you wish the students to learn. "Activity" is the way the students go about acquiring this knowledge. As an example, one might want the students to learn chemical and physical properties of acids and bases. The activity carried out by the students could range from passive listening to a lecture, listening and answering some questions, reading and answering questions, watching a film, carrying out experiments, or some combination of these.

Motivation of students has its difficulties for besides having to consider the five factors listed earlier, it must be remembered that no matter how much one might like to think of a class as a single entity, it is actually a group of individuals with many real differences.

Tyler[14] lists the following as "major dimensions of individual differences": intelligence, talents, personality, interests and values, cognitive style, and school achievement. These in turn are due to the relationships that have developed between the genetic and environmental influences surrounding each individual.

There are two ideas here worth considering before proceeding. One is the fact that a teacher, particularly at a secondary school level, can do little to remove these differences. They exist, and as a result many attempts at motivation are always likely to leave some students uninterested.

The second idea is the listing of school achievement as separate from intelligence (when the latter is measured by intelligence tests). Studies indicate that intelligence tests are more highly correlated with

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[14] L.E. Tyler, The Psychology of Human Differences (New York: Appleton-Century Crofts, 1965), p. vii.

scores on standard achievement tests than they are with grades given by teachers.

In her summary of these studies, Tyler concedes that this lack of correlation is probably partly because intelligence measures are not independent enough of learned information. She thinks this is also partly because "teachers do judge pupils on characteristics other than mastery of the subject matter"[15]. She observed that "Cooperativeness, agreeableness, persistance, and willingness to work could all be involved"[16].

This means that one must not lose sight of the teacher's differences from some or all of the students with respect to interests and values. This can affect the teacher's behaviour when interacting with the students.

As an example, in a study by Keddie[17] it was found that teachers viewed those students who would not easily accept the teachers' views and classifications as the poorer students, while those who did accept the teachers' statements without question, and who were viewed by the teachers as like themselves, were considered the better students.

One can see that due to the variation in individuals and five different factors of motivation to consider the possible combinations can lead to many different results.

It is very difficult to set up situations which everyone will like, so even establishing a proper attitude in everyone is not as easy

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[15] Ibid., p. 109.

[16] Ibid., p. 115.

[17] N. Keddie, "Classroom Knowledge", Knowledge and Control, ed. Michael Young (Toronto: Collier-MacMillan, 1971), p. 151-156.

as it sounds at first. Either or both the social and academic standards set by the teacher can be at variance with the expectations of some of the students and a conflict can arise which will interfere with the environment of the other students.

Academic and social standards must be carefully thought out so as to be realistic. A teacher must look at his own behaviour to see if he is being overly critical or demanding and is adding to, if not causing, an already anxious state in students.

Bruner compares the student activities of "coping and defending"[18], and although he believes that everyone does both to some degree, concludes that some learning problems of children are due to such an extreme case of defending themselves from criticism that they fail to learn. This unconscious fear of trying can only come from anxiety, but the causes may be due to the home situations and out of the teacher's control.

Also, levels of readiness differ. As a class enters a new grade at the start of a session, it is assumed that the students all have the ability (i.e. readiness) to do the work the course entails, since at least the majority of the members have been promoted from the previous grade. This assumption is a fallacy, but it is one with which both students and teachers are stuck. Remedial aid may help a few, but even there, requests for aid can take as long as the whole school year before they are processed and the assistance even starts.

A study by Learned and Wood cited in Tyler[19] makes it quite clear that "differences (in achievement) within a grade are far greater than

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[18] J.S. Bruner, Toward a Theory of Instruction (New York: Norton & Co., 1966), Chapter 7.

[19] L. Tyler, p. 107.

the average differences between grades". In fact, roughly a quarter of high school seniors knew more (i.e. could answer more questions on a "general culture" test) than students with two years at college[20].

Even if one still assumes that all the students can handle the learning, it becomes an increasingly doubtful assumption to think that they all can do it at the same rate and with the same amount of work.

If some are going to need more time and expend more effort, it brings us back to their willingness and aspiration. Do they see a reason for this extra time and effort?

Being successful at learning or doing some task can raise the aspiration levels of students and establish an attitude that is satisfying. But everyone's goals are not the same. Too many small successes can cause unrealistic goals. Unrealistic goals will eventually be recognized as unattainable and result in frustration and disturbance in class.

Attempts to increase aspiration levels by lowering teacher expectations and making the students' work easier have the potential to cause the reverse of what is wanted. Instead of feeling successful, the students can recognize that the work is "easy" and show even less effort than they did before. This result is certainly less likely to occur if the desire to learn the material is strong.

Even increasing the willingness factor of motivation is not without its problems, but unfortunately the usual attempt by most teachers to increase this willingness to learn is by reference to future goals rather than immediate uses. As was pointed out earlier, this approach

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[20] Ibid., p. 104.

of future goals is almost without effect.

If motivation has problems, then what about reinforcement methods?

Everyone agrees that reinforcement (both positive and negative) is best if it is immediate and precise.

Skinner,[21] for example, contends that immediacy is virtually impossible on written tests. He also feels that teacher's comments on papers are also liable to be weak and imprecise.

Reinforcement of verbal answers can be done quickly by a teacher, but personal comments may be interpreted incorrectly and result in complaints of favouritism and hostility. Also, the teacher may respond to and hence reinforce the attention-getter. Unfortunately for teachers it is easy to adopt a habitual type of response to answers. Yet Sorenson[22] claims that studies indicate that teachers who use the same words of praise repeatedly do not have as great an affect as those who were spontaneous.

Skinner feels that the best reinforcers are successes, for then "he (the student) not only knows, he knows that he knows and is reinforced accordingly."[23] Similarly, on a written paper the true reinforcers are largely automatic; "a sentence comes out right, it says something interesting, it fits another sentence."[24]

Sorenson's view is similar to this. "Successful learning reduces

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[21] B.F. Skinner, The Technology of Teaching (New York: Appleton-Century Crofts, 1968), p. 152.

[22] Sorenson, p. 421.

[23] Skinner, p. 156.

[24] Skinner, p. 160.

the tension involved in accomplishing it, and the satisfaction which comes just from the release of tension positively reinforces or strengthens the learning."[25]

Bruner[26] considers the will to learn to be an intrinsic motivator that finds its reward in its own exercise. He feels that external reinforcement may get a particular act going, and even lead to its repetition, but it does not foster a long course of learning.

With secondary school students this natural curiosity is more jaded and dulled by previous exposure to topics and years of the apparently arbitrary and meaningless goals of teachers. This means that it can not be given the same emphasis in secondary school, but his observations do parallel the beliefs of Sorenson and Skinner that success is its own reward and is important in creating confidence and an increasing willingness to keep going.

Negative or aversive control in the form of criticism or punishment is used by teachers and is not completely detrimental[27], but the fault with this negative reinforcement is the fact that the student is not taught the correct response; instead he is punished for the wrong one. This wrong behaviour may be extinguished but it may be replaced by another wrong one rather than the correct one.

Thus with reinforcement differences in student personalities and goals may reduce its effectiveness. Large class sizes and rotating classes will also reduce the likelihood of a teacher knowing the students well enough to match the proper immediate reinforcing response to an

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[25]Sorenson, p. 421.

[26]Bruner, pp. 127-128.

[27]Sorenson, p. 425.

individual.

Even if internal reinforcement does have a greater effect than external, both face the same problem. There can be no reinforcement until the student has made the correct action. If there is no willingness to make this action, then the actions will be few and far between.

Every graduate of a teacher training institution will have been exposed to the factors of motivation, activity and reinforcement to some degree, but after finding that motivation and reinforcement are not easy to use effectively, most secondary school teachers eventually concentrate on the activity portion of the learning steps and call that motivation.

I must state that I am not basing this opinion on any formal study done by myself or anyone else, but rather on my own experience and thirteen years of observing what fellow teachers do and say, and the topics at Professional Development meetings. Words like "motivation" and "reinforcement" are occasionally used, but it is the activities that get almost all the consideration and emphasis. Even when the word reinforcement is used, it is frequently meant as a synonym for drill. The usual "motivation" is derived from trying to stimulate the students with an interesting content presentation (i.e. some activity) instead of trying to interest them in the original reason <u>for</u> the content.

In searching for a methodology for teaching, teachers are most likely to look for a model on which to base their own behaviour, and the usual choice is the behaviour of their own teachers.

However, teachers are part of the small percentage of the population who were successful enough in school to remain there until graduation from a university. This really means then that they are

choosing (unconsciously) a model that "works" for only part of the population and thinking that it will be successful with everyone to whom they teach.

Although R. Gagné discusses college instructors in his article, I feel that he has reached the same conclusions as I have made about secondary school teachers. As he puts it, "Most college instructors set about their initial task of teaching courses by using a model derived from their own college experiences; in other words, they try to emulate their own professors.....it is obvious that the new instructor is perpetuating many traditions. It is also true that this traditional system may be said to 'work'.....because it has worked for him and for most of his fellow students."[28]

This approach gives little thought to motivation or reinforcement. Even consideration of alternative activities to make the content more interesting is often based on the assumption that because the teacher finds it interesting, so will the majority of students.

This action is understandable when one considers that the teachers' own strengths lie in their knowledge of the course content, and there is no outside encouragement to search elsewhere for a solution to problems.

Much of this emphasis may be due to the influence of Dewey, for although his ideas have been altered by many, one concept appears to have been retained. That is the importance of student interest. Unfortunately this has been interpreted incorrectly. Dewey states that, ".....learning through interest is not merely an easy or lazy way of

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[28] R. Gagné, "Learning Theory, Educational Media and Individualized Instruction", The Curriculum: Context, Design and Development, ed. R. Hooper (Edinburgh: Oliver & Boyd, 1973), pp. 299-300.

acquiring a certain amount of knowledge; it is a way of calling into play the full powers of effort and determination of the learner."[29]

Because of their pre-occupation with course content, the above statement or others like it have been taken to mean that teachers should try to make each activity interesting instead of making the content interesting.

For example, if the content is the digestive system, little or no attention is given to interesting the students in the system itself or finding reasons why they might benefit from learning about it. Instead, questions, diagrams, etc. are likely to be interspersed with several experiments, dissections or films, so that the activities of learning will be varied and, it is hoped, ensure that the class is not bored with the entire topic.

However, when emphasis is placed on activity the students can mechanically go through the motions of working or studying, thereby giving the appearance of motivation, but without learning very much. Therefore even a well thought-out sequence of content material is just not good enough by itself, nor are interesting activities the final answer.

Thus, besides looking for interesting activities, teachers should be looking for content that is applicable and useful to the students themselves, and telling them of this usefulness.

Dewey is also of the opinion that "pupils are little interested in the distant future.....Education as preparation for life is only

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[29] S. Curtis and M. Boultwood, A Short History of Educational Ideas (London: University Tutorial Press Ltd., 1965), p. 479.

significant if it refers to life now and in the immediate future."[30]

If the students can see uses for the content they are learning they will be interested even if the activity portion of the learning is very passive. In fact, in a comparison of teaching methods, listening to lectures, which is the least active of all possible activities by a student, was found to be equal to any other method for learning factual information.[31]

A typical question frequently asked by students is of the type, "Why are we taking this?" which shows that the student can see no relevance of the topic to his life.

This kind of question is a source of frustration for teachers for if the latter are interested they cannot understand a student's lack of interest. The feeling grows that the students must be lazy or not intelligent enough, or some combination of both. Any carrot held out is that of future use....neglecting Dewey's assertion that students are primarily concerned about the immediate future.

With all the references in studies to the motivational power of usefulness and immediacy it is apparent that these are the factors on which teachers and course planners should be concentrating.

The next two chapters give an example of how one can place more emphasis on usefulness, as a way to stimulate willingness to learn. This does not imply that a teacher should ignore other means of motivation, reinforcement or any interesting activities that might be available. Rather, increased willingness should in turn increase

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[30] Curtis and Boultwood, p. 479.

[31] N. Gage and D. Berliner, Educational Psychology (Chicago: Rand McNally, 1979), p. 445.

attention and cooperation and make all the other factors involved in learning more available for use.

CHAPTER THREE

GENETICS AND MUTATIONS

Having stated my educational goals and the reasons for them in the previous chapters it is now necessary to show how these goals might be realized in practice.

This entire teaching unit of genetics and heredity is subdivided into what I shall call four topics: genetics (the basic laws and patterns of inheritance); mutations (exceptions to the expected patterns); defects and counselling (knowledge of birth defects, possible treatments and the need for professional guidance); and ethics (the moral issues raised by responses to the threat or existence of genetic defects).

Although there is a flow or development of ideas from one topic to the next, for the purpose of this project I shall describe the genetics-mutation theme in this chapter and the defect-counselling theme in the next. Thus for the most part, this chapter will deal with content that is familiar to someone who has taught biology, while the content of chapter four will be less so.

The basic content of the unit is that required by the Ministry guideline. The pertinent pages of the document are duplicated in Appendix 4.

Any alterations from the expected basic content are in the number of items studied and these arise as a result of adding content pertaining to genetic defects, counselling and ethics, and organizing the early material with these additions in mind.

Since the goal is to present the content in a way that the students

will see as useful and pertaining to themselves, these alterations will not necessitate any loss in the development of "scientific literacy" or the development of basic concepts needed for future academic study.

Throughout this example unit I see no need to go into detail about some of the factual content that is included (e.g. how to do punnett squares or what words mean), unless it seems necessary to clarify what I am proposing, or to describe areas that I think should get extra attention. Such detail is not needed by readers to weigh the merits of my approach to motivation, and it is already familiar to biology teachers who may wish to try the unit as described.

There are three changes in this topic from that proposed in the Ministry Guideline. These are: the order of topics, the stress on human genetics, and the introduction of pedigree studies.

As Appendix 4 shows, self-replication of DNA is included in Unit II of the Ministry Guideline along with mitosis, but meiosis is part of Unit VI. The roles of DNA and RNA in protein formation are not mentioned at all. Instead of this organization, I teach the structure and roles of DNA and RNA all in the unit containing mitosis, and teach meiosis and the rest of Unit VI(a) along with Ministry Guideline Unit III, 2(f) as a separate unit concerning reproduction. Both of these are done before genetics.

Similarly, mutations are part of the Guideline Unit VII concerning evolution, and are shifted to this genetics unit, leaving the remainder of the evolution content for later.

These changes are summarized on the following page as Table 3.1, with only the pertinent sections shown.

The table shows that these changes are in the order of topics, so

TABLE 3.1

CHANGES IN ORDER AND CONTENT FROM THAT SHOWN IN MINISTRY GUIDELINE.

Unit II Cells

7. Mitosis

includes replication of DNA and explanation of action of chromosomes and genes.

add: roles of DNA and RNA in protein formation (new).

Unit III Organisms

2(f) Reproductive Systems

add: Meiosis (from unit VI b)

add: Duplication of individual (from unit VI a)

add: Start of Life (from unit VI c)

Unit VII Evolution

remove: recent ideas of mutation (b)

Unit VI Heredity and Variation

remove: (a) duplication, (b) start of life, and (c) meiosis, all of which are now in 2(f) above.

add: recent ideas of mutation (from unit VII)

add: pedigrees (new)

add: human birth defects and counselling (new)

add: ethics of counselling (new)

reorganize this unit into the four topics as previously described.

the Ministry requirements of content and intent are still met. Thus, for any reference to information about meiosis, DNA or other units in the course, it can be assumed during the genetics unit that the items have already been taught earlier in this course, and the students will have that background information.

To emphasize human genetics, discussions, questions, etc. should pertain to human characteristics. If any examples or material concerning plants or other animals seem necessary they can be slanted to illustrate human interest in selective breeding.

This may sound too anthropocentric, for the laws of inheritance exist for all organisms, but it is important that the students understand that these laws do affect humans.

Statements in genetic and genetic counselling literature about the public's knowledge are usually similar to that given by Margaret Thompson, Professor of Medical Genetics at the University of Toronto. "The amount of ignorance displayed unfortunately, by the young people who come to our counselling clinic is appalling and saddening"[32]. And yet, as has been noted elsewhere, "As more infectious and nutritional disorders yield to treatment and control, more of the total health-care burden will be represented by those chronic disorders that have a genetic component... diseases such as coronary heart disease, cancer, diabetes...in which a persons genetic constitution plays a contributing role".[33]

Human genetics is also much more interesting than non-human genetics

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[32] M.W. Thompson, "Human Genetics as a scientific discipline and a social concern" (paper presented at the STAO conference, Toronto, November 4, 1978).

[33] J.D. McInerney, Human Genetics Education: Background and Rationale, The BSCS Journal, Vol. II, (2), (Boulder, Col.: April, 1979), pp. 2-3.

to the students, for they would rather talk and learn about themselves than about peas or fruit flies. Thus the first step in increasing the willingness level of the students to learn about genetics is to make it apply to them personally whenever possible.

The second step is to make it look (and I hope prove) useful to them in the near future.

The third major change from the "expected" content is the introduction (as shown in Table 3.1) of a family history to detect the presence or type (i.e. recessive, X-linked, etc.) of a genetic trait.

GENETICS

To introduce the topic of genetics I begin with the continuum of human traits that is present today, as represented by Figure 3.1, on the following page.

By itself, this figure is not informative but by using primarily a lecture approach with some recitation, this introduction should give the points that follow the figure. This recitation, as defined by Bellack et al[34] consists of the following steps:

> "1. The teacher provides structuring, briefly formulating the topic or issue to be discussed.
> 2. The teacher solicits a response or asks a question of one or more students.
> 3. A student responds or answers the question.
> 4. The teacher reacts to the student's answer."

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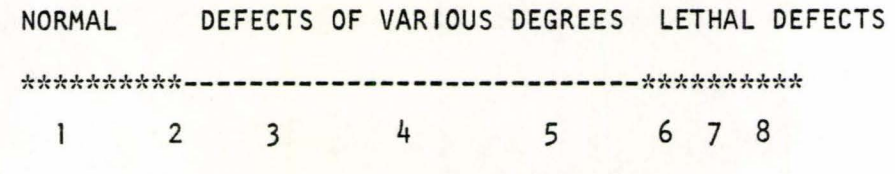
[34] N.L. Gage, D.C. Berliner, Educational Psychology (second edition), (Rand McNally, 1979), pp. 625-26.

FIGURE 3.1

CONTINUUM OF HUMAN TRAITS



position	description
1.	healthy (no apparent defects)
2.	minor defects (e.g. allergy)
3.	minor disorders (e.g. colour blindness)
4 & 5.	increasing severity of disorders (e.g. mental retardation, heart defects, limb defects)
6.	lives for years but dies young (e.g. Huntington's Chorea)
7.	dies at birth or very young (e.g. Tay-Sachs)
8.	dies before birth.

The points to be brought out during this introduction are:

a) some traits are controlled by single genes (i.e. qualitative), some by genes in combination (i.e. gene interactions) while the environment plays a significant role in the expression of polygenic systems.

b) qualitative traits result in "all-or-nothing" variation, much of which society accepts as normal (e.g. two sexes, attached ear lobes).

c) interactions between genes and/or polygenic systems both cause quantitative traits where many genes each contribute a small part to the trait. These result in a range of variation, much of which society accepts as normal (e.g. height, strength).

d) traits outside the common range or common varieties are seen as abnormal even if they are not considered defects (e.g. exceptional

height).

e) defects can be considered to be traits that are so abnormal that they cause death or deviation from the normal mode of living in a particular society. These too show both variation and range in size (e.g. Tay-Sachs, hemophilia).

f) the potential inherited variation (including ranges and defects) exists in the would-be parents and it is possible to calculate the probability of occurence for some of these traits.

I chose this type of figure by way of introduction because the students are already accustomed to horizontal lines like this to show gradual changes (i.e. electro-magnetic spectra, chemical periods).

The use of body stature as a trait showing range is one to which students can easily relate. They are surrounded by classmates who they see as being taller or shorter than themselves and yet still consider both these others and themselves to be normal. It is also better to start by acknowledging that characteristics cannot be divorced from environmental influences, and that all characteristics are not controlled by only one gene.

The purpose here is to start with knowledge the students already have, and then to extend this with two basic ideas. These are: the range in physical and mental ability is not often controlled by single genes in an on-or-off manner, and; defects can be considered to be further cases of the same ranges and variations, and therefore capable of the same methods of study.

The students are not expected to learn this continuum or the names and/or symptoms of disorders at this time.

The last part of the introduction will be to provide the students

with an overview of what topics the unit includes and to give them orally a brief description of each. Emphasis is on the desire to predict serious birth defects that might occur.

Although most of this introduction would be by lecture, student participation might suggest alternative examples to those listed in the points above. This participation may reveal information regarding the knowledge that students already have about genetics and genetic defects, although this is not the main purpose.

The actual objectives of this topic are in the genetics section of Appendix 1, but the basis of these is the ability to do punnett squares and work out and use pedigree studies.

Therefore almost all the remainder of this topic can be done by presenting the class with prepared stencils containing questions involving each type of situation (e.g. mono- and dihybrid crosses, co-dominance, multiple alleles, X-linkage and combinations of these) for which the students should be able to do a punnett square, a pedigree, or a combination of both and arrive at a conclusion.

One at a time the teacher will present an example question(s) of each of these situation types (e.g. monohybrid cross) and demonstrate on the blackboard how to solve each type. After each example the students will practice that type of question using the questions from the stencil before the teacher goes on to the next.

In the course of demonstrating solutions, it will be necessary to give and/or explain the terminology in the questions and answers. This should be done <u>as it is needed</u> and not beforehand.

It is good organization to have the students learn, as a preliminary exercise, all the word meanings they will need so that the

terminology can be used later in explanations. However, it is not good teaching. Learning the word meanings this way does not show where each applies and this lessens the likelihood of the students understanding either the meaning or any explanation in which it is used.

Therefore, the terminology should only be explained as each word or expression arises.

Showing examples and assigning questions certainly is not any different than what many teachers now do. The difference for motivational purposes, is in the questions themselves.

First, the questions deal with humans. Second, traits due to genetic defects are used far more than is common in texts, and these will aid in showing the students the usefulness of being able to make predictions about offspring. Third, some questions are asked that involve the students in a personal manner. What will <u>their</u> children be like? This should strengthen the idea that this topic contains information that is useful to themselves.

Unfortunately, it is difficult to devise questions that use defects as the trait(s) and are also personal without introducing fictional spouses, or ailments.

Personal questions if real could be embarrassing. On the other hand, if the defect does not exist in that student, then there is really no question.

For this reason it is necessary to use "normal" traits like eye colour in some questions and examples, and assume any required simplification.

According to Milunsky[35], questions involving hair and eye colour

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[35]A. Milunsky, <u>Know Your Genes</u>, (New York: Avon Books, 1977), p. 308.

usually cannot actually be answered for these are more complex than a question assumes. As an introduction to genetics, however, I think one should be free to make some simplification in order to bring out the main point or points of interest.

This is done in the first example used (i.e. pollen allergy) and in question 12 that follows.

The questions on the following pages are those that the students would receive on a stencil for this topic.

GENETICS QUESTIONS

In all questions in which only the phenotypes of the individuals are given, it may be necessary to try more than one genotype.

For example, curly hair (C) is dominant over straight hair (c), and a parent who has the phenotype of straight hair must have the genotype, cc. However, a parent who has curly hair can be either CC or Cc. Therefore, for the second parent both genotypes must be used to make predictions about their offspring.

1. A brachydactylous man marries a normal woman. They eventually have both normal and brachydactylous offspring. This is an autosomal dominant trait.
   (a) What are the possible genotypes of the man and the woman?
   (b) Prepare a punnett square (PS) to show all the possible genotypes of the offspring.
   (c) What fraction of their offspring will probably be brachydactylous?

2. If your spouse turns out to be heterozygous for curly hair, determine what fraction of your children will probably have straight hair.

3. (a) A man and a woman know that they are both carriers (i.e. heterozygotes) of galactosemia. What are the odds that none of their offspring will have the defect?
   (b) The same man and woman are also both carriers of cystic fibrosis (on a different chromosome) which they do not know. Prepare a PS to determine all the genotypes of the offspring they might produce, including galactosemia, and state what fraction of each phenotype will be produced.

4.* Give the gene content of the different kinds of eggs produced by a woman whose genotype is JjKkLl? (Kimball #3 p. 275).

5. Select one member of the opposite sex from the class and work with them to decide the probability of producing each of the blood types, if you two were to have children. (If you do not know your own type, males use A, and females use B.)

6. Suppose two newborn babies were accidentally mixed up in the hospital. In an effort to determine the parents of each baby the following blood types were determined.

   Baby 1 = Type O Mrs. Brown = Type B Mrs. Smith = B
   Baby 2 = Type A Mr. Brown = Type AB Mr. Smith = A

   Determine which baby belongs to each set of parents.

7.* Distinguish between multiple factors and multiple alleles. (Kimball, #1, p. 330).

8.* Some genes in humans are known to have a multiple (or "pleiotropic") effect on the phenotype. Does this constitute an invalidation of the one-gene-one-enzyme theory? (See page 328 of Kimball.) (Suzuki, #18, p. 251).

9.* A phenotypically normal man in his early twenties learns that his father has developed Huntington's chorea.

(a) What is the probability that he will develop the symptoms himself in later life?
(b) What is the probability that his son will develop the symptoms in later life? (Suzuki, #4, p. 17).

10.* A woman who has Muscular Dystrophy in her family including her brother, wishes to have children.

(a) What is the risk of her being a carrier?
(b) What is the risk of any of her children having the disease?
(c) Without knowing if she is a carrier, what is the probability of passing this disease to a son?
(d) If she should have a son who has the ailment, then what is the probability of the next son being affected? (Adapted from Suzuki #1, p. 41).

11. Males do part (a), and females do part (b).

(a) If you were to marry a woman known to be heterozygous for hemophilia, and whose father was colour blind even though she has normal vision, what phenotypic ratios can you expect in your sons? your daughters?

(b) If you were to marry a man with red-green colour blindness and brown tooth enamel, what phenotypic ratios can you expect in your sons? your daughters? (Brown tooth enamel is a dominant X-linked trait).

12. A very close friend has come to you for advice. He has suspected that his wife has been having "an affair" for some time with a neighbour, and now that she has produced a son, your friend does not believe himself to be the true father. He gives you the following information.

Himself: brown eyes, free ear lobes, no hemophilia, type A blood.
Son: blue eyes, free ear lobes, no hemophilia, type O blood.
Wife: brown eyes, attached ear lobes, known carrier of hemophilia, type B blood.
Neighbour: blue eyes, free ear lobes, no hemophilia, blood type unknown.

Draw any reasonable conclusions possible, and justify them.

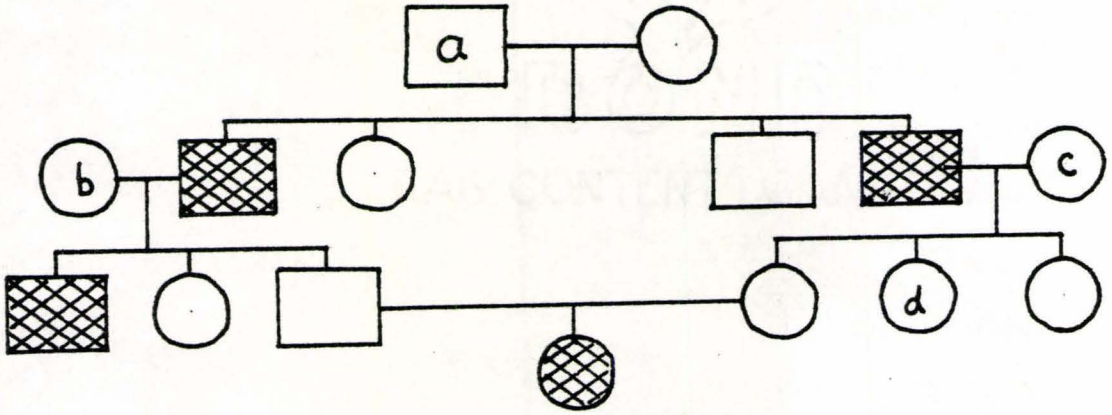
Brown eyes are dominant over blue (autosomal). Free ear lobes are dominant over attached (autosomal).

13.* Achondroplasia is a form of dwarfism inherited as a simple monogenic trait. Two achondrolastic dwarfs in a circus marry and have a dwarf child. Later they have a second child who is normal.

(a) Is achondroplasia produced by a recessive or dominant allele?
(b) What is the probability that their next child will be normal? dwarf? (Suzuki #5, p. 17).

14.* (a) What kind of inheritance for the trait T is illustrated in the following pedigree diagram?

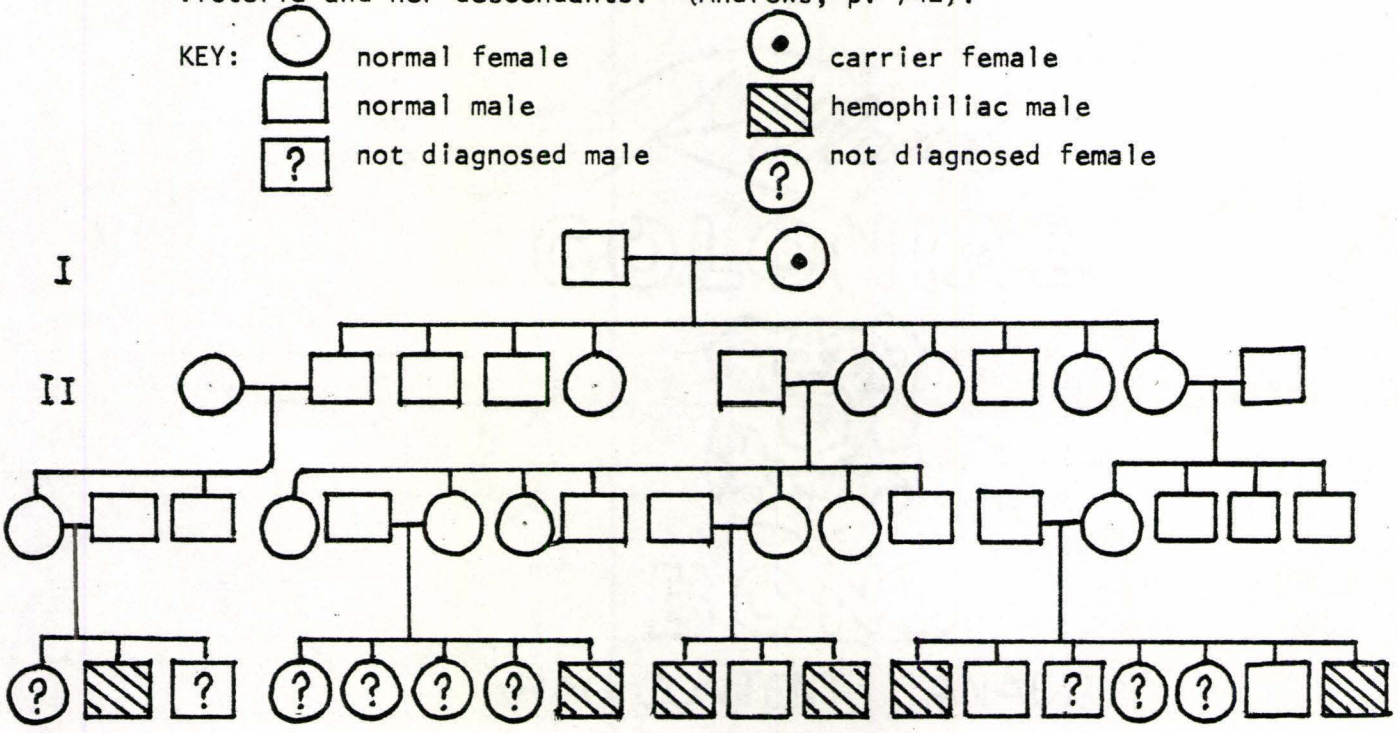
1) autosomal dominant 2) autosomal recessive
3) X-linked dominant 4) X-linked recessive.



(b) What are the genotypes of a, b, c and d? (Galbraith, #15, p. 754).

15.* The following figure represents Queen Victoria's pedigree but the code marks indicating who were hemophiliacs, who were carriers, and who were normal have not been shown for generations II and III.

Work backwards from generation IV, marking in the intermediate generations until you have a reasonable pattern to connect Queen Victoria and her descendants. (Andrews, p. 742).



* Indicates the question was taken or adapted from one of the following texts:

W.A. Andrews et al. Biological Science An Introductory Study, (Scarborough: Prentice-Hall), 1980.

D. Galbraith and D. Wilson, Biological Science: Principles and Patterns of Life (3rd ed.), (Toronto: Holt, Rinehart and Winston), 1978.

J.W. Kimball, Biology (Don Mills: Addison-Wesley), 1978.

D.T. Suzuki, An Introduction to Genetic Analysis, (San Francisco: W.H. Freeman and Company), 1976.

The questions shown cover all the types of situations (i.e. mono-, dihybrid, etc.) that can be included in Mendelian genetics.

The examples of each type of situation that would be done as demonstrations should be simple and yield straight forward answers.

The first example of such a situation I would use is the following question, written on the blackboard:

> A man who is heterozygous for pollen allergy and a woman who is homozygous for no allergy plan to have children. If the allergy is dominant over no allergy, then what fraction of their children will probably also be allergic to pollen?

There are words here that must obviously be defined and the definitions written down as the solution progresses. After completion of a punnett square, the genotypes of the offspring can also be compared to show the meanings of geno- and phenotype.

This approach of example(s), terminology, explanation and practice questions would be continued until all the situations to be encountered in the exercises are eventually covered.

Where more than just the ability to do punnett squares or pedigrees is required, explanations are to be given as the need arises. This involves descriptions of sex chromosomes prior to X-linkage, describing karyotype analysis, and work on, or explanations about probability. Some or all of these explanations can be done by students reading from the text, although short lectures can be as informative and more directed to the topic objectives.

Experience has shown that completion of punnett squares is a mechanical exercise that does not trouble students who do the practice.

Although the type of questions proposed should encourage that practice, the work should not be too repetetive or boredom may offset the willingness you are trying to encourage.

It may be necessary of course, to make up more questions or examples if these are needed. Similarly, it may not be necessary in any one year to use all the questions on the stencil. Many of these questions require more work than a typical text book question for the students are given only phenotypes here, and so must do more than one punnett square to see what offspring can be produced. This is also more realistic for most individuals do not know their genotypes.

Eventually Mendel's laws can be stated and written out formally, and his work with peas described to explain expressions like "Mendelian ratio". Similarly, a summary should be given at the end to emphasize that no matter what the trait(s) under consideration, for single gene traits, the same ratios always show up. For example, in monohybrid crosses, two heterozygotes theoretically always yield a 3:1 ratio; a dominant homozygote and anything else always yield an all: 0 ratio, etc.

The real problems in genetics are establishing what traits are dominant (or recessive) as well as monogenic, and the genotype of the parents--not in doing the punnett squares. These problems are far greater when humans are involved in the studies for there is little or no control of human mate selection or environmental influences such as diet, pollutants and activities. If this "complication" of limited study of humans has not already arisen through student questions in the course of the topic it must be brought out to direct the students towards using family history information.

The first work with pedigrees can be done as an experiment

involving each student's family. Using paper treated with phenylthiocarbamine (PTC), each student can trace the ability to taste (or not taste) the chemical through their immediate family using guide sheets provided for this purpose. (See Figure 3.2 on the following page.) Application of this method to determine whether a trait is dominant or recessive will demonstrate how this approach can be used with other traits.

If no PTC paper is available the same experiment can be done using the trait of "tongue rolling" for both this and the ability to taste PTC are dominant.

A second point to bring out during the post-lab discussion is the ability to use this same method of tracing through a family to predict the presence of a particular gene in a particular family member. It is this use that is important for predicting the presence of genes that might lead to genetic birth defects.

As more work on pedigrees is included in the counselling topic only two more questions for students are given in the work for this topic.

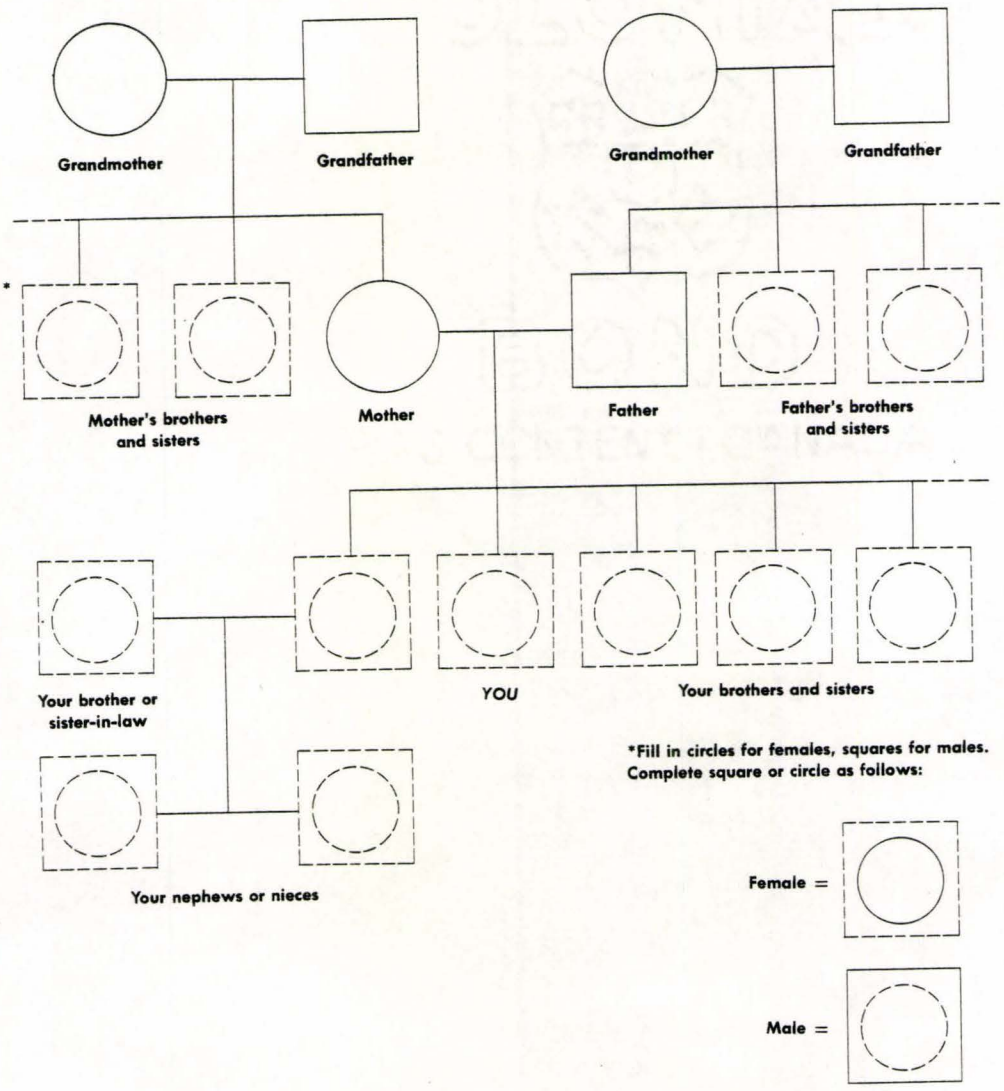
Although genetic <u>defects</u> are not directly part of this topic, some of these have been used in problems as some of the traits in question. Information concerning these about dominance, and autosomal or X-linked nature which is needed for the questions is to be provided on hand-out sheets. Copies of these are found in Appendix 3. Other information is also included but that will not be used in this topic.

It should be noted that mutations, mosaics, and variable penetrance have not been introduced at this point. Complications such as these are better left until the students have gained confidence and ability with more straight forward situations.

I believe that the use of genetic defects in some questions,

FIGURE 3.2

WORKSHEET ON THE ABILITY TO TASTE PTC[36]



On the basis of the information collected above is the ability to taste P.T.C. paper transmitted as a dominant or recessive gene?

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[36] P. Abramoff, R. Thomson, Investigations of Cells and Organisms, (Englewood Cliffs: Prentice-Hall, 1968), p. 358.

personal traits in others, and the wording of some questions to make them more like problems, will all combine to show the students that this topic is useful to society in general and even more important for motivation, to themselves both in the distant and the very near future. This being the case, their willingness to learn and understand both the material studied and any further implications it might lead to, should increase.

Once the "mechanics" of doing punnett squares and pedigrees are learned the students should have no great difficulty with the given questions or similar ones on tests or exams. This success should raise their aspiration levels. When combined with their willingness to learn material that appears useful, the result should be a "proper" classroom attitude for they should like what they are doing.

Because a great deal of the work is done at their seats, the teacher can move about and assist and/or provide verbal reinforcement to students as each is needed. Both of these should reduce anxiety that might be present. Readiness does not apply much in this topic of genetics. It is new information, or you are assuming that it is new, so failure to understand earlier units in the course should not have any effect, except with meiosis. That brings up my last point in this topic.

Two areas where students can have difficulty at first are with probability and determining what genes are in the parental gametes. These two areas must be given extra attention.

To determine genes in gametes, I believe the starting point is with meiosis. This topic can be reviewed quickly with sketches to illustrate assortment and segregation of genes on the chromosomes, similar to Figure 3.3 on the next page. These sketches can then be referred to when examples are done, or questions taken up, by changing the

FIGURE 3.3

DISTRIBUTION OF GENES DURING MEIOSIS

The illustration uses only two pair of chromosomes, each with 2 alleles, A and a, and B and b. (Not to scale).

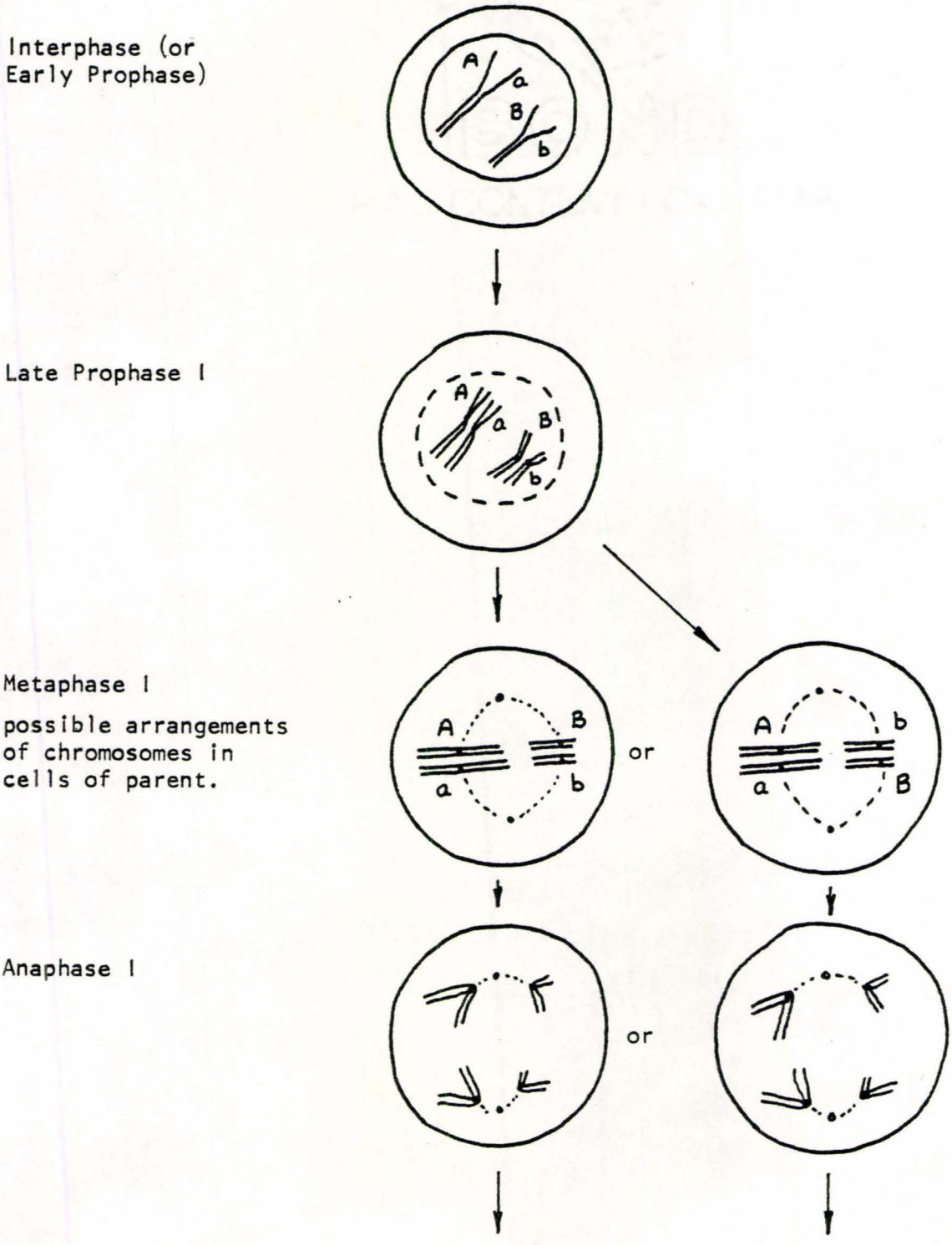
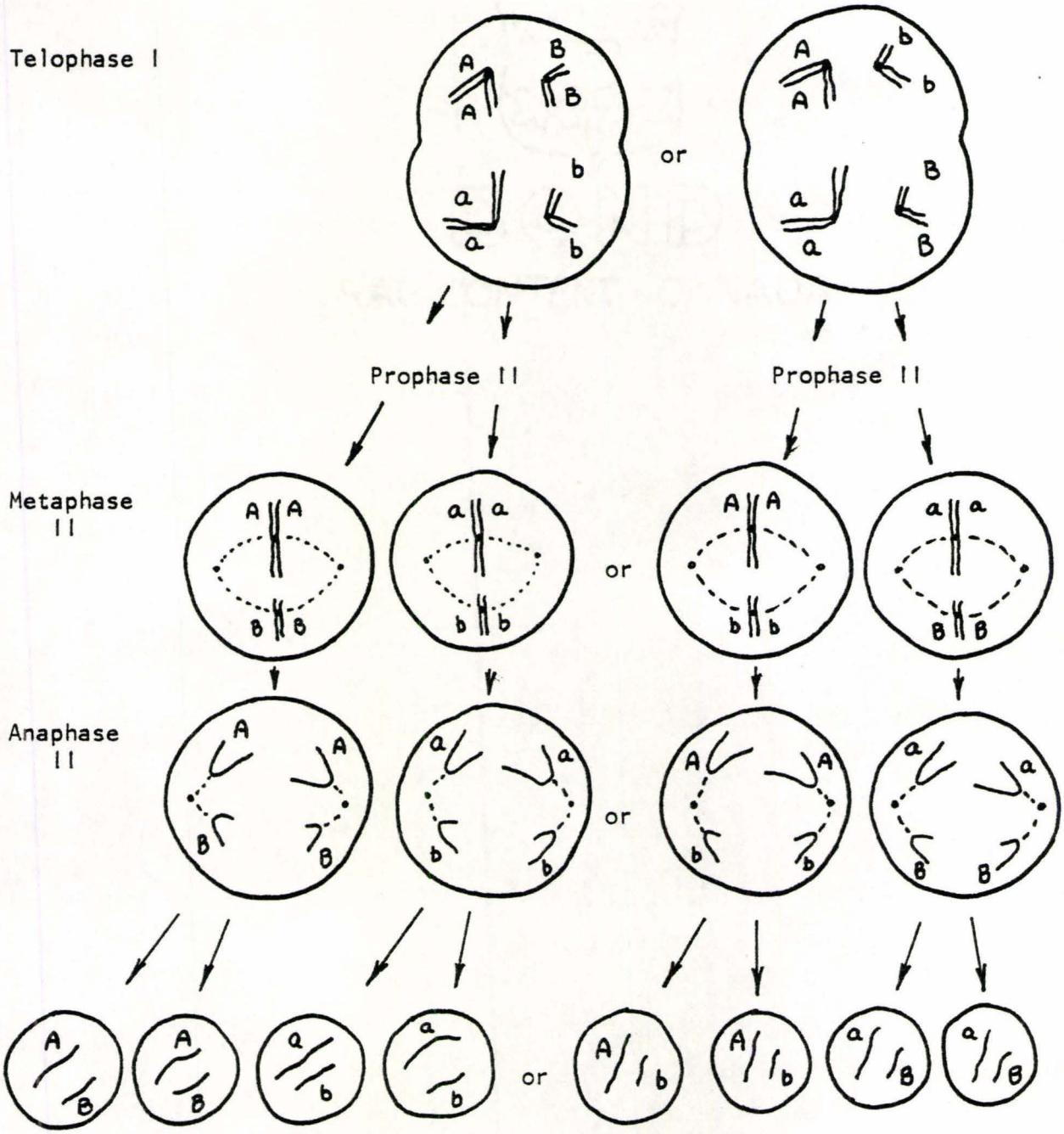


FIGURE 3.3 (Cont'd.)



There are 4 different arrangements of chromatids possible in the gametes of a parent.

letters representing the genes to correspond to the trait(s) in the situation under study.

These references can be discontinued when everyone in the class is observed to be setting up punnett squares properly. Student's work should be checked for this.

During probability work it must be stressed that a ratio in the offspring of, for example, 3 to 1: normal to affected, does not guarantee that only one or always one of the actual offspring will have that trait. Milunsky[37] claims that some parents may think that because their first child is already affected, they will be safe in their future pregnancies. Such an erroneous belief would stop them from seeking professional counselling.

It is unnecessary for the students to become experts at probability questions, but they must understand that the ratios determined in a punnett square or empirical observations are simply chances.

It is advisable to have an experiment ready whereby students can observe probability so that this exercise can then be used with any individuals who seem to need it, or if necessary, with the whole class.

Such an exercise could require the students to roll sugar cubes with a given number of faces marked, or to toss a coin(s) and tabulate and examine the results of a number of rolls or tosses. An example of such an exercise using coins is found in the intermediate level text by Erwin et al[38].

A better approach in that it is faster and more directed is that

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[37] A. Milunsky, Know Your Genes, (New York: Avon Books, 1979), p. 139.

[38] G.W. Erwin et al., Methods of Science Today 4, (Toronto: Clarke, Irwin & Co. Ltd., 1979), pp. 186-188.

used by Welch et al[39], in which no experiment is done. Rather, the text starts by giving probabilities of common events requiring explanations, and gradually works to the point where students calculate the probability and explain their reasoning.

Examples of the types of questions are: explaining why the chance of drawing a spade in a single draw from a full deck is 1/4, or predicting the chance that the next girl passing will have brown hair and be slim if one girl in every two has brown hair and one girl in three is slim.

By using this approach, much of the work on probability can be done orally with the class, with only a few examples worked out and explained on the blackboard for their future reference.

The main ideas needed and brought out are:

(i) the result of one trial of a chance event does not affect the results of later trials of the same event.

(ii) the chance that two independent events will occur together is the multiplication product of their chances of occurring separately.

However, the weakness of only giving odds and not guarantees must be stressed for this is important in the counselling and ethics topics to follow.

The result of this work should be a successful accumulation of the knowledge and skills required by the topic. The content starts from very general knowledge of variation which the students already have, and grows to include new knowledge. The approach should provide a teacher with time to give individual reinforcement and assistance and provide the students with proper levels of all five motivational factors.

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[39] C.A. Welch et al., Biological Science: Molecules To Man (Boston: Houghton-Mifflin Co. 1973), pp. 317-322.

The innovative feature of the study of this topic, however, is the presentation of the material in problems that show the students the potential for using the knowledge to make personal decisions (i.e. decisions that affect themselves) in the future.

Because the unit is not yet complete, and the information will continue to be developed, there should also be the feeling on the part of the students that further knowledge can change the potential use into an actual use, and this should increase their willingness to learn both this topic and the ones following.

A schedule of items to be covered and the expected time requirements is suggested in Appendix 2 for each of the topics. However, in actual practice I would start the next topic in mutations before all the questions on genetics are completed, and therefore would overlap the two.

MUTATIONS

The roles of the X and Y chromosomes in sex determination were previously explained in the first topic, so an interesting way to begin any work on mutations is with several examples of sexual abnormalities resulting from aneuploidy.

As study of this topic progresses one will eventually distinguish these chromosomal aberrations from point mutations.

By presenting the major symptoms of Turner's, Kleinfelter's and "superfemale" syndromes and then relating these to the chromosome complement present in each case, one can involve the students, through question and answer, in determining how these particular complements arose.

The goal is not to have the students learn specific segregations.

Up to this point in the unit, both punnett squares and pedigrees have been used to trace and/or predict the appearance of variations that already exist in individuals. The goal of this topic, starting with this lesson, is to explain how these phenotypic variations arose in the first place, and how they may occur again.

Experience has shown that students are very interested in sexual aneuploids and it is by taking advantage of this high interest that one can lead them deeper into the topic of mutations.

The rest of the introduction can be done by lecture, for the initial information is factual and a lecture provides an opportunity to get quickly into the topic (i.e. meanings of mutation, causes and results to the genes, etc.). Moreover, the lecture allows one to show the overall organization of the topic.

A lecture also allows one to relate this topic to the last one by pointing out that if abnormally functioning genes or some rearrangement of chromosomal segments occurs, these will be passed along just the same as if they were normal.

The actual content of this introduction should only be the meanings of mutation, mutant, congenital, for these are needed immediately, and lists of the presently known causes of mutations, and the four cell types that can be altered. These will be explained shortly.

The objectives shown in Appendix 1 list the factual content to be covered throughout this topic, but it is appropriate to clarify the overall concepts that hold these facts together. One concept is that all mutations do not result in changes in phenotype, for this depends on the type of cell in which the mutation occurs. As will be shown, it is necessary therefore to relate these changes to cell types.

A second concept is how the actual source of a mutation causes the changes in the DNA, and thus the changes in the cell genotype. At this grade level it is unnecessary to go into great detail about these DNA alterations.

There are different ways to define mutation and different ways to classify and therefore count mutational sources.

Because of the concepts to be developed in this topic it is necessary to choose a definition of mutation that includes somatic mutations. As Suzuki says, these "...are 'potentially' but not necessarily heritable from generation to generation."[40] Such a definition would be "[a mutation is]...any change in the DNA that has the potential to cause a change in the functions of the cell".

This definition allows one to consider the four types of cells: somatic, germ, gamete, and for the purposes of this unit, embryonic. Because of their lack of differentiation and their rapid division, these latter can be considered to be germ cells. However, the number of daughter cells affected by a mutation in an embryo will be far larger than the number that arise from a mutation in a germ cell of an adult. Thus, for the purposes of discussing the dangers and results of mutations it is better to treat embryonic cells as a separate type.

To classify the causes of mutations I have chosen a mutagen approach rather than look at the results (i.e. morphological, biochemical), for this is easier to relate to exposure and/or avoidance. Thus, three causes to be studied are radiation, chemicals and viruses. Besides these

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[40] D. Suzuki, A. Griffiths, An Introduction to Genetic Analysis (San Francisco: W.H. Freeman and Company, 1976), p. 199.

three there are also spontaneous point mutations, and chromosomal aberrations, some of which can be related to age. Although there are theories that the latter may be caused by previous exposures to the first three listed (i.e. radiation, etc.) or combinations of these, these are unproven as yet. Those related to age are important for the next topic for when counselling is considered, it is the age of the patient that is known, and more easily considered than previous exposures.

Avoidance of repeated lecture-explanations by the teacher requires the use of questions for student research and answer. Examples of these are on the next page. The answers to these questions should provide the factual information needed to meet most of the student objectives.

MUTATION TOPIC

PART A

1. Radiation can cause mutations and genetic disease. Describe the change(s) in the structure of DNA resulting from radiation that might lead to these disorders.

   What are the "safe" limits of exposure? Why is there difficulty in setting true safe limits?

   What are possible sources of radiation exposure, and to which of these are you exposed? How can you reduce your exposure?

2. Viruses have been linked to congenital and mutational changes. Describe how these changes may be brought about by viral action.

   What viruses are known or suspected to cause disorders in humans due to genetic change?

   Is it possible to reduce the likelihood that you will suffer from any such genetic disorder?

3. Many drugs and chemicals have been linked to congenital, mutational and cancerous disorders. Describe how these types of chemicals bring about the changes in DNA structure responsible for these disorders.

   List at least four chemicals known to cause these disorders and state which type of disorder (listed above) they cause.

   Name at least four of these chemicals to which you are exposed. Is it possible to reduce your exposure?

4. Chromosome breakage can alter the messages contained in the DNA. Assuming that this occurs in meiosis, describe how these breaks can occur, and the results with respect to distribution of genetic material.

   Sketch a graph of the observed frequency of Down's Syndrome versus maternal age and suggest a possible explanation for this relationship.

PART B

In all of the questions in this part, make the following assumptions: (i) the mutation does not result in cancer; (ii) any altered gene is one that is active. That is, if the gene responsible for insulin production is mutated in the muscle cells of the leg, there will be no effect for that gene is not operative or active in those particular cells anyway.

5. You have been exposed to a mutagen and it has caused mutations in several of your somatic cells.

(a) Describe the effect on your body if this mutation is (i) dominant (ii) recessive.

(b) Describe the effect on your future offspring if this mutation is dominant.

6. You have been exposed to a mutagen and it has caused mutations in several of your germ cells (e.g. several cells in your bone marrow will now produce defective red blood cells).

(a) Describe the effect on your body if this mutation is (i) dominant (ii) recessive.

(b) Describe the effect on your future offspring if this mutation is dominant.

7. You have been exposed to a mutagen and it has caused mutations in several of your gametes.

(a) Describe the effect on your body if this mutation is (i) dominant (ii) recessive.

(b) Assuming that one of the mutated gametes is used in the production of one of your offspring, describe the effect on this offspring if the mutation is (i) dominant (ii) recessive.

8. You or your spouse have been exposed to a mutagen while pregnant and it has caused mutations in several cells of the embryo or fetus.

(a) If the embryo is only a few weeks old, what effect will the mutation have if the mutation is (i) dominant (ii) recessive?

(b) If the fetus is about 8 months developed, describe the effect on it if the mutation is dominant and again if it is recessive.

9. Seriously harmful mutations rarely exist in the gene pool of a population as a dominant trait. Explain why such a mutation is much more likely to be recessive and find or make up a reasonable example to aid in showing this phenomenon.

PART C

10. Previous mutations in a population have caused what are known as "inborn errors of metabolism". The variety of errors has in turn resulted in many ailments present today. What is the error in each of the following ailments? alcaptonuria, phenylketonuria (PKU), and sickle-cell anemia.

11.* More than 10 000 new molecules are synthesized each year. Many of these could be mutagenic. How would you readily screen large numbers of compounds for their mutagenicity? (Suzuki, #10, p. 219).

12.* In humans only autosomal trisomics for chromosomes 13, 18 or 21 survive until birth. However, with the sex chromosomes, all of the

following chromosome abnormalities have been observed: XXY, XXX, X, XYY, XXXX, and XXXY.

(a) State what sex each of the above individuals would be.

(b) How many Barr bodies would you expect to find in the cells of each of the individuals above? (adapted from Kimball, #9, 10, p. 294.)

13. What is the source of "reverse transcriptase", and why is its existence surprising, dangerous and useful?

* indicates the question was taken or adapted from one of the following texts.

J.W. Kimball, Biology (Don Mills: Addison-Wesley), 1978.

D.T. Suzuki, An Introduction to Genetic Analysis, (San Francisco: W.H. Freeman and Co.), 1976.

Of course it would be best if each student did every question but because of the time needed to do these, and a possible shortage of resource material, a compromise is needed. I suggest that the students work in pairs, with each pair being assigned only two questions; one from part A and one from B. All students will answer part C.

In order for all the information to be collected by all students it must be passed around. To do this each pair can present their findings on one of the questions to the rest of the class via a verbal and blackboard presentation. All students' written answers can be graded with or without a portion of the total being allocated to the classroom presentation.

If this grading is done before the classroom presentations, possible additions, deletions or alternate wordings can be pointed out, so that the rest of the class receives the needed information in a concise manner.

Mixed throughout these student presentations, at appropriate times, short lectures and explanations can be given by the teacher to complete the objectives (see Appendix 1) of the topic. Similarly, any recent news items concerning mutations or the threat of these can be pointed out for possible discussion.

At this point the students have received sufficient background to go on to further academic study in genetics in accordance with the Ministry requirements.

Because mutations can lead to diversity, larger gene pools, adaptation and eventually to evolution of a species, as well as cancer, death or disfigurement, one must consider carefully how far into each of these possible results one wants to delve. I believe that to maintain student interest in genetics, as little divergence as possible into

adaptations and/or cancer should be done at this stage of the unit. It is the use of the knowledge by the students (i.e. avoiding or predicting birth defects) in the near future that arouses an interest. To move from the flow of ideas relating genetics, mutations and birth defects to discuss adaptations and evolution would weaken that flow and thereby weaken any interest.

If information on these other issues did arise during discussions or student questions, it is still only those student objectives listed in Appendix 1 that would be used for tests and examinations.

At some later time in the course, if one wishes to pursue the ideas of adaptation and evolution, the knowledge the students have acquired about mutations in this unit will provide the basis they will need.

The text currently in use by the grade 13 biology class is Biology (4th edition) by J.W. Kimball. An alternative is Biological Science (3rd edition) by Galbraith and Wilson. The material in this topic is found as follows:

	Kimball	Galbraith
gene mutations	pp. 308-09, 598-99	pp. 764-6, 713
chromosomal mutations	pp. 283-87	pp. 767-68, 694
cell types	p. 308	

As can be seen from the page numbers, not only is the material provided in little detail, it is scattered due to the authors relating it to other material.

This means that any teacher intending to go into more depth than these texts do, must be prepared to provide other texts or articles from either the libraries or personal sources. These are easily found, for

all texts on genetics that I have seen have more than enough information on mutations. Besides those listed in the bibliography, other possible references are:

Levine, R.P. Genetics. Toronto: Holt, Rinehart and Winston, 1966.

McKusick, V.A. Human Genetics. Englewood Cliffs, New Jersey: Prentice-Hall, 1964.

Pedder, I.J. and E.G. Wynne. Genetics: A Basic Guide. New York: W.W. Norton & Company, 1972.

Watson, J.D. Molecular Biology of the Gene (3rd Ed.). Don Mills: W.A. Benhamin Co., 1976.

Winchester, A.M. Human Genetics (2nd Ed.). Columbus Ohio: Charles E. Merrill Pub., 1975.

The content presented to the students throughout this topic is of two types with respect to its motivational power. That concerning cell types and changes in DNA is only useful for those who select future study in biology. For the present it is included so that the students can understand the rest of the mutation topic, but this will not make it useful in the eyes of most students. As explained in chapter one, it is inevitable to have some of this type of information in most topics, and this should not cause a problem provided study of the topic does not stop at this point but continues until it includes useful material.

The rest of the topic, such as recognizing exposures to and avoiding the sources of mutations, should be clearly emphasized as being useful to everyone, for it is. The students can start watching for these sources the day that they learn of them. It is this material that will help to balance out the "non-useful" material and maintain the motivational factor of willingness by preventing the entire topic from taking on the

appearance of being strictly for background knowledge.

Readiness for this topic depends upon knowledge of DNA and its role (studied earlier in the course) but because no great deal of detail is required or expected in describing any DNA alterations this should not pose a problem.

Anxiety might increase when the students first find they have to research and answer questions using books other than their own texts, but if it is clear that you are not expecting detailed answers, and check their work giving hints, guidance and reinforcement as they work, this anxiety should disappear.

In general, I do not expect aspiration or attitudes to change during the study of this topic. The content and the expected objectives are quite factual, and intricate problems are difficult to set up due to the limited nature of the material. As always, individual differences may alter these motivators slightly for some students are very happy to memorize facts while others are not. This is such a short topic, however, that neither of these types of students should be greatly affected.

A short lecture should finish the topic by verbally summarizing what the students have learned. At the same time it can provide two points. The students themselves may have had or will have mutations which they can pass to their offspring. Also, most genetic ailments are the result of previous mutations already in the population that are being passed from generation to generation.

By ending with a summary lecture such as this, during which genetic ailments are related to mutations, one is ready to progress to the next topic of genetic disorders and counselling.

CHAPTER FOUR

GENETIC DEFECTS, COUNSELLING AND ETHICS

There are two reasons for including genetic defects, counselling and ethics in a secondary school course and both arise from the topics' potential usefulness for practical decisions.

The first is that already stated. That is, they allow the teacher to show an important application of the material which the students are expected to learn about genetics and mutations. Since this increases their willingness to learn such material, the topics should be studied for their instrumental value in enhancing student motivation.

The second reason has nothing to do with motivation. It is based on the belief that this is information that everyone should have because it can affect the quality of life, and the way to reach most people is to start with students at early grade levels. Although I am suggesting that it be taught to grade thirteen students I still believe in its general importance and hope that anyone who is not convinced about its motivational value will nevertheless consider its inclusion in future biology courses at all levels.

Citizens should know more about genetic ailments and counselling to aid in decision making. A representative passage from the literature is the following: "...at least 90% of the people in the United States, West Germany, and other Western countries who really need counseling do not receive it."[41] For those who do receive counselling, a needs

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[41] A. Milunsky, Know Your Genes (New York: Avon Books, 1977), p. 134.

assessment project carried out by the March of Dimes says, "Young people mostly lack specific knowledge of genetic disorders, since even most modern biology courses in high school and college say little about them. The genetic counseling literature repeatedly emphasizes that such a lack of understanding constitutes a major obstacle to effective counseling and the subsequent reduction in the incidence of birth defects."[42]

A look at some facts about genetic disease may be needed to show the importance of knowledge of these disorders to the individual and society.

A copy of the following facts will be given to the students but more will be said about that later.

1. 36 percent of all spontaneous abortions (i.e. 100 000/yr) are due to genetic disease.[43]
2. 40 percent of all infant mortality is due to genetic disease.[44]
3. 80 percent (or 12 x 10^6) of Americans hospitalized each year are the result of gene or chromosomal birth defects.[45]
4. 30 percent of the patients admitted to pediatric hospitals in North America have diseases that can be traced to genetic causes.[46]
5. 10 to 12 percent of the population has enzyme abnormalities which

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[42] Biological Sciences Curriculum Study, Guidelines for Educational Priorities and Curricular Innovations in the Areas of Human and Medical Genetics (Boulder, Col.: 1978), p. 23.

[43] National Institute of Health, What Are The Facts About Genetic Disease? (Bethesda, Maryland: DNEW Publishers, not dated), p. 6.

[44] Ibid., p. 21.

[45] Ibid., p. 6.

[46] D. Suzuki, A. Griffiths, p. 4.

make them likely to react adversely to one or more commonly used drugs.[47]

6. Chromosomal abnormalities occur at a rate of 1/200 live births. This is over 20 000/year in the U.S.A.[48]
7. There are currently 2000 disorders known to be genetically linked, and this list is growing at the rate of 75 to 100/year.[49]
8. About 3 to 4 in every 100 babies are born with major birth defects.[50]
9. There are over 6 million persons in the U.S.A. who for either hereditary or congenital reasons are mentally retarded.[51]
10. At least 1 in 10 individuals has, or will develop, some disorder that has been inherited.[52]

GENETIC AILMENTS AND COUNSELLING

Besides deciding what specific objectives the students should meet, there is also a decision needed about how much material concerning genetic ailments they should read. There is little in the way of texts or guidelines to which one can refer for other opinions on this.

Much of the literature seen by myself tends to deal with the role of the clinician which is far more detailed than what is needed by the

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[47] National Institute of Health, p. 9.

[48] A. Milunsky, The prenatal Diagnosis of Hereditary Disorders (Springfield, Ill., Charles C. Thomas, 1973), p. 136.

[49] National Institute of Health, p. 9.

[50] A. Milunsky, Know Your Genes (New York, Avon Books, 1977), p. 2.

[51] Ibid., p. 2.

[52] Ibid., p. 1.

students.

As a self-made guide to determine the specific behavioural objectives (see Appendix 1), what and how many examples of disorders to provide, and what other reading or lectures would be required, I developed the general aims listed below.

1. The students should know that there is a range of ailments (e.g. polydactylism to Tay-Sachs) and what some of these are. The students must not be left with either the impression that genetic ailments are all insignificant or that they are all large and disastrous.
2. Students should know that although new ailments may occur in a family at any time from mutational causes, many already exist in the family. Determining what these are would involve studying the family history.
3. Students should be able to apply mendelian ratios to known cases, but should recognize that most cases are not that simple so that some predictions are quite weak. They should also be aware that some disorders are brought out or intensified by environmental factors.
4. Students should know that some ailments can be tested for prenatally, others postnatally and some not at all. They should realize that a decision to use tests would involve a knowledge of amniocentesis and ultrasound procedures and their associated risks.

Students should also know that:

5. some ailments can be treated and others cannot, and of what these treatments might consist,
6. some people are in need of counselling more than others are, and what factors make this so,
7. there are professional counsellors available who should be consulted if a potential risk is thought to exist (i.e. medical geneticists

specializing in this field rather than family physicians or non-medical geneticists),

8. there are emotional, social and economic problems that can face either or both the parents and the afflicted child.

Although this may look like a lot of information, it can be condensed into very few behavioural objectives, as shown in Appendix 1.

Much of this information has already been given to the students via the stencils given out during the genetics topic. Samples are in Appendix 3. This served the purpose then, of allowing variety by using different defects in the questions as well as providing a reason for trying to make predictions about offspring.

The criteria for selecting the defects to be studied was so that they and the accompanying data about them would be used in the study of this topic to show variation in size of defects, treatments, possible tests, as well as the source (i.e. autosomal, X-linked, etc.).

It is not necessary that the students learn about large numbers of these ailments with their corresponding tests and/or treatments for much of this specific information will be forgotten in the future for this is not information they will use. Rather, the general aim is to have students recall that differences do exist in treatments, size of effects, and so on. Thus there is no reason to have the students learn more than six disorders; 3 minor and 3 major. What constitutes major and minor is open to personal opinion, but most would agree that disorders like Lesch-Nyhan, Tay-Sachs, etc. are major, while ones like polydactyly or colour blindness are minor, due to the degree to which "normal" life is upset.

I feel that requiring this number now is sufficient to provide the

basis for remembering for many years that variation does exist so that all genetic defects will not be stereotyped, nor will professional counselling be ignored. For the present, it also allows the students some individuality in selecting defects that "appeal" to them, and consequently should help increase interest in the field as a whole.

To introduce this topic, one should begin by presenting the class with the same statistics about genetic defects that were given earlier in this chapter. The verbal emphasis to accompany this is that these defects are not rare or isolated cases and that they could appear in the offspring of the students. The aim is again to instil a personal interest in this topic.

The next step is to have the students read through the stencils they were given earlier in the genetics topic, and list all the treatments and tests for disorders given there. This involves them in summarizing the information they will need presently.

What should come next, then, is a discussion of who should seek genetic counselling, and a listing of these points as they are elicited. Since anyone with a family history of a genetic ailment, or a pattern that appears to be due to the genetic makeup of their family, should seek counselling, an assignment should immediately follow this.

The given assignment requires students to set up his or her own pedigree chart by working backwards in their own family. However, instead of just dates, what should be recorded are the causes of death, any serious illnesses and any early deaths, known for each member they can trace. (See question 1 of the questions for topic 3 that follow.)

Besides giving several days to do this, some extra explanation should be given. One point is that you expect them to be honest in both

their answers, and how far back they can go in family lines. What they are looking for is a pattern that might mean something, and it will benefit them to get information now for it will get harder to find this information as times goes on and more and more relatives die.

Because you are expecting them to be honest, the results of this may prove embarrassing for some, either after it is done, or even before, if they know what it will show. With this in mind a little lecture about not feeling guilty or embarassed about genetic disorders is in order, but it should also be guaranteed that no one will see the pedigree chart except the teacher and depending on whether they are to be evaluated, maybe not even him/her.

A genetic counsellor at McMaster University stated that one of the two main problems facing patients was not knowing enough about their family history.[53] It is hoped that this type of assignment will aid in helping to solve that problem.

Subsequent lessons would involve two reading assignments and accompanying questions pertaining to ultrasound and amniocentesis, short lectures on the topics of environmental influence and predictions involving multi-factorial inheritance, and the role of a genetic counsellor. Interspersed through these would be questions on the various items as shown on pages 67 and 68.

Diabetes mellitus, atherosclerosis and spinal bifida are good examples to use to show the influence of environment with respect to the onset or severity of some ailments. Although there is a genetic cause to each of these, there are other contributing factors. Diabetes is

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[53] Interview with Jamie Dianda, Counsellor, McMaster University Genetics Programme, Hamilton, Ontario, 1981 07 07.

influenced by diet, pregnancy, age and geography, atherosclerosis by diet, sex (hormones), and environmental stresses (smoking, job), and spinal bifida by maternal age, season of birth (or gestation) and geography. (Further details of these three ailments can be found in Milunsky's book, in chapters 21, 18, and 5 respectively.)

Even if the geography factor is a reflection of the number of carriers in an area this should be considered in making predictions for some ailments. For example, a Canadian descended from immigrants from Northern Ireland has a higher probability of spinal bifida than one descended from immigrants from southern England. As Milunsky[54] notes, "....7 in 1000 births reduce to 2 per 1000 in southern England. ....the Irish in Boston have kept their higher risk of spine and brain defects."

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[54]A. Milunsky, Know Your Genes (New York: Avon Books, 1977), p. 62.

QUESTIONS FOR TOPIC 3. GENETIC COUNSELLING AND DEFECTS

1. Using your own family, prepare a pedigree chart showing all members of your family for as far back as you can trace. Rather than trace any one ailment, however, try to determine and then mark in known carriers or victims of all genetic diseases. Also mark in any family members who died at any age from unknown diseases, or who are known to have suffered with an ailment throughout their lives.

   What you are to look for are any and all patterns that might become apparent. Be honest and get as much information as you can. The results may be important to you.

   Depending on your findings it may be impossible to use any one code, so write in your findings about particular members if this method is easier.

2.* A man and a woman have normal vision. They have three offspring all of whom marry people with normal vision. These three offspring and their children have the following phenotypes.

   (a) a son with red-green colour blindness who in turn has a daughter with normal vision.
   (b) a daughter who has three sons with normal vision.
   (c) a daughter who has one son with normal vision and one red-green colour blind son.

   Draw the pedigree for the family, indicating the affected people and the carriers. (Kimball, p. 328).

3. Read section 18.9 starting on page 328 of your text, and list the three principles that can alter how the genotype is expressed. Give an example of each of the principles by thinking of one of your own. Do not use the ones in the text.

4. Describe the structure and the function of an operon. What is the value of these units in terms of feed-back?

5.* Consider the following table concerning two recessive metabolic diseases found in humans. The figures represent units of enzyme activity and show the range found.

Disease	Patients	Parents of Patients	Normal Individuals
Acatalasemia (enzyme catalase)	0	1.2-2.7	4.3-6.2
Galactosemia (enzyme gal-1-P uridyl transferase)	0-6	9-30	25-40

This information is available for many metabolic genetic diseases.

(a) Of what use is it to a genetic counsellor?
(b) Indicate any sources of ambiguity.
(c) Reevaluate the concept of dominance in the light of such data. (Suzuki, p. 252).

ULTRASOUND IN MEDICAL DIAGNOSIS[55]

1. What is the basis of this type of examination?
2. For what two reasons do the authors consider the risks of ultrasound to be "innocuous"?
3. What is the problem caused by the strong reflections from bone or gas?
4. At what points in the body do sound waves bounce back?
5. What are the uses of ultrasound examinations in obstetrics?

GENETIC AMNIOCENTESIS[56]

1. What is the advantage cited for prenatal diagnosis?
2. What percentage of the women who could benefit from amniocentesis actually have the procedure performed?
3. When are fluid samples usually taken?
4. What role does sonography play in amniocentesis?
5. How much time is required to culture fetal cells?
6. What are the four types of disorders that are detectable?
7. Describe the procedure for this type of examination.
8. What are the two "major hazards" from the procedure?
9. (a) What percentages of fetal loss arose from amniocentesis subjects and from controls? (b) What is the percentage of other symptoms?

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[55] G. Dewey, P. Wells, "Ultrasound in Medical Diagnosis", Scientific American (May, 1978), pp. 98-112.

[56] F. Fuchs, "Genetic Amniocentesis", Scientific American (June, 1980), pp. 47-53.

10. What conditions does the author consider should be weighed when determining the risk-benefit ratio of amniocentesis?

* indicates the question was taken or adapted from one of the following texts.

J.W. Kimball, Biology (Don Mills: Addison-Wesley), 1978.

D.T. Suzuki, An Introduction To Genetic Analysis, (San Francisco: W.H. Freeman and Company), 1976.

Spinal bifida is also a good example of a polygenic disorder (i.e. one environmentally influenced) to discuss predicted occurences of defects. Although these vary slightly between races and geographical locations, the following chart[57] makes a good summary.

Conditions	Risk of having
both parents normal	an affected child is 0.003%
both parents normal but they have had one affected child	a second affected child is 3-5%
one parent is affected	an affected child is 3-5%
both parents normal but they have had 2 affected children	a third affected child is 8-12%
both parents normal but they have had 3 affected children	another affected child is about 25%

These risks are based on average empirical results and not on Mendelian calculations so the risks to an individual could differ from these. A counsellor, however, with no other information to go by would have to use these average predictions.

This is a good place to have the class read the article "Who Shall Be Born"[58], which describes the feelings of expectant parents undergoing AFP tests for spinal bifida, and their decisions about what to do. This then leads to a discussion of the role of a genetic counsellor.

The role of a counsellor is to obtain facts and then to use these to make a prediction, if possible, on the odds of an event such as a birth defect. Besides this, the students should be told that the counsellor will not make decisions for them. With this in mind, a discussion about risks should follow, while trying to minimize the discussion of

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[57] A. Milunsky, p. 107.

[58] G. Chedd, "Who Shall Be Born", Science 81, II, No. 1 (Jan.-Feb. 1981), pp. 32-40.

ethical matters as yet.

Assuming that the probability or risk is known, the students must be made to consider whether they each believe this to be a significant degree of probability. This will vary due to individual differences in personalities, background exposures, future plans, etc., and is further complicated by the seriousness of the defect in question. Two individuals might both opt for an abortion if the risk of their offspring having Tay-Sachs syndrome is even 5% because it is a fatal defect. But if the ailment is Down's syndrome and the risk is still 5%, one of these same individuals may still opt for an abortion, while the other might be willing to forego abortion altogether (no matter what the risk) for the defect is not immediately fatal. These decisions of course are based on the individual's values and how they weigh those values against each other. Thus, although there may be a correct decision for an action for each individual and situation, there is no one correct decision that can be applied to all.

The "correct" decision concerning the two alternatives must be the one which will please that particular person the most, or more realistically, displease him or her the least, for it is he or she that must live with the results. Although the emphasis here should be considering risks as high or low, this overlaps with the ethics topic to follow, and is a good tie-in to that topic.

During study of this entire topic, and the next, it is essential that the teacher refrain from presenting him or herself as an authority on all possible disorders, and from giving actual counselling advice.

Because the factual requirements are not long or complex, and the application of these facts in decision making is based on the student's

personal value system, the motivational factors of aspiration, attitude and willingness should all remain high.

The big factor in the topic is willingness. I believe the students will want to know more about the frequency and symptoms of many disorders. As these are learned they will also want to learn about the tests, etc. that are available. As long as the time devoted to this topic is kept short, which it is, the interest will remain high.

The only previous material needed in the way of readiness is making predictions using pedigrees and in some cases punnett squares. These items they should all be able to do, so they should have neither problems nor anxiety due to this.

Any anxiety that arises will be for two reasons. The first is about actual future birth defects and not about learning the material. It may even increase the willingness to learn the material.

The second source may be due to a student having birth defects himself or in his family that he does not want discussed. It is important therefore to state (and restate) that you shall not discuss anyone's personal problems in class. As for the family history assignment, I think that other than looking quickly to see if they are done, I would not even read them. They are doing this for themselves--not for you the teacher. This should be clearly stated beforehand.

Thus, the proper motivational factors are all present, and the presentation of the facts and ideas has been varied by using lectures, reading, answering questions and discussions. All of the work leads the students to think about how the information can be applied to themselves. The variation should prevent any boredom that might arise from the continual use of any one particular method of instruction.

At the same time, consideration of risk and the role of a counsellor leads directly into an examination of the ethics of a decision.

ETHICS

The title of this topic is somewhat misleading for this is not meant to teach ethics to the students.

Except for the process of genetic screening which would be explained, and some arguments for and against it presented in the classroom recitation method, the majority of this topic is to be done by presenting problem situations.

These problems are intended to start the students thinking about how they might face genetic disorders in their children or what decisions they might make as a result of information acquired from a genetic counsellor. It is to be hoped that by considering some arguments themselves, they will become more tolerant of people who make different decisions than they would make provided that the decision is based on valid reasons.

During the interview with the genetic counsellor J. Dianda, cited earlier, she stated that the decisions involved were the second main problem faced by patients. Having been given medical and/or statistical information, they then have difficulty weighing the pros and cons of a particular action (or non-action) as it applies to themselves. Many of these parents or parents-to-be have never before considered their social responsibility or how a situation might affect their lives, and in many cases have little time to do so before a decision must be made.

A discussion of these problems will not eliminate the difficulty of making a decision later, should the need arise. It may lead to

better decisions, however, by providing some guidance as to balancing the priorities in personal values with actions and results.

Discussions also allow the students to hear pros and cons that as individuals they might not think about either now or in the future.

In teaching this topic, one can obtain little or no assistance from general texts. Moral problems (particularly in medicine) are issues that have been avoided by science authors[59] although there are some that can give a teacher some background information to help develop the topic.

The main concerns are the use of genetic screening, and the use of abortion, although questions can also include determining probabilities or risks (using punnett squares and pedigrees), consideration of the severity of the disorder, and possible treatments. These are all factors that would be weighed in a true situation.

Although at times it may be necessary to provide information which the students overlooked (so as to give a complete picture of a given situation) it is essential that in all cases teachers keep their views to themselves and thus avoid influencing the students. This is difficult to do. In a situation where the probability of an occurence is 3 to 1, views can be given subtely in how the risk is stated. Is it given as a 1 in 4 chance of a deformity or as 3 chances in 4 of a normal birth? If repeated often enough the students will infer what the teacher thinks.

Making a correct decision in a situation depends upon adequate knowledge but it also depends on the personality of the person(s) in question. Thus, to begin, I would point out to the students that in a

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[59] P.D. Hurd, "The Historical/Philosophical Background on Education in Human Genetics in the United Stated", BSCS Journal, 1(1), (February 1978), p. 3-8.

given situation there are four possibilities using the following as an example.

Four women (A to D) are told the babies they are carrying have Down's syndrome, and the results are as follows:

A. she aborts and later regrets the decision to do so.

B. she aborts and is satisfied.

C. she has the child and is satisfied with the decision.

D. she has the child and later regrets this choice.

It is obvious that in cases B and C the women made choices acceptable to themselves even though they made opposite choices. There is no one proper solution for each made the "correct" choice for herself.

In cases A and D the women either did not have all the facts or did not consider the facts carefully enough in view of the effects on their lives.

There is also the possibility that these women may have even made the right choice but lack information about the defect or themselves and do not know they made the right choice. No matter how unsatisfied they are with their decisions they could be even more unsatisfied with the other choice if they were living with it. This is a case of wanting a happy or a good decision, whereas in reality all the choices are bad, with some worse than others.

Thus, in any discussion in class or in a real situation what the students should try to do is look at all the facts that might have an effect, consider the effects by weighing them and then present these. The overall purpose of this topic is thus to show them what factual information they might want or need, and to have them practise weighing alternatives.

The concept of a "correct" decision is a decision that the person can face after it is made, and not suffer guilt feelings or regret. It may not be a "nice" decision where everyone lives happily ever after, but one that the person can accept as the best of two bad alternatives, and feel that in view of their life goals and values it was the only realistic choice.

The "questions" done by the students in this topic are of two types: those involving situations that imitate a personal decision, and those of a more social nature that involve some of the aspects of genetic screening. This latter type requires a lesson to explain the meaning and reasons for genetic screening. At the same time one can present the apparent success with Tay-Sachs and Thalassemia and the problems that have arisen with PKU and sickle cell anemia.

The actual "questions" that are to be given to the students are provided below. For both the personal decision type questions and the screening topic questions, these are to be solved in groups of three (or four for a large class). One group would present its collective arguments for one action or attitude, while a second group would present those of the opposite view.

I believe this debate approach works even if a member does not agree with the side for which he is arguing. In fact, besides each person hearing all the points that can be brought out, such a person may see the other opinion better. This in turn may help in developing a tolerance to differing opinions, even if it is not an objective to be measured.

To avoid embarrassment or anxiety due to saying how they would behave under certain conditions, it is better to assign the students which view point to take and argue for, with clear instructions to present

it as best they can. Thus, they can find themselves arguing for a view point that they do not agree with. However, no one will know for sure whether it is really their opinion or not.

QUESTIONS CONCERNING TOPIC IV

PART A: Genetic Screening

Instructions: The statements following present a view or opinion on various aspects of screening. Some students will be assigned to present arguments to support this view while others will be assigned to attack this view and give arguments to support a contrary position. Whether you agree personally with the side you are assigned, do the best you can to come up with valid arguments to support it.

1. Genetic screening should be voluntary and not mandatory.

2. If the parents of a child with a genetic defect refused testing, or refused an abortion after learning of the disorder's presence, then they should receive no financial aid to help them meet the cost of this defect.

3. If the parents of a child with a genetic defect were tested and told that the chance of their offspring being normal were 99%, but it was still born with the defect, they should still not receive financial aid.

4. Once screening for ailments becomes common, any victim that still suffers a genetic ailment will be viewed by society as some sort of outcast.

5. Anyone who elects to undergo tests to detect potential birth defects should pay most if not all of the costs of these tests themselves.

PART B

Instructions: In each of the situations following a question is asked. However, depending on your own personal views, you may want additional information before answering it. If this is the case, state what else you would want to know, and how that information would affect your decision.

1. Due to their wish to have a home and job security, a couple has delayed having children while they both worked. Now that they are both willing for the wife to stay home she has become pregnant. However, she will be age 34 at the birth of the baby. Should they have an amniocentesis done?

2. A man and a woman have been trying to have children for 6 years and finally the wife has become pregnant. The reason for the problem is not known, but it is not due to miscarriages. A few weeks after the news of the pregnancy they learned that the wife's mother is a carrier of muscular dystrophy.

   What action should they take? If the baby is male, should they opt for an abortion?

3. You (or your male spouse) are aged 25 and you have just learned that your (or his) father has Huntington's chorea.

   What will be your decision should a pregnancy develop in your own family? Explain.

While the debates are going on each of the remaining class members can picture themselves as a "patient" hearing these arguments, and then make a decision based on what goes best with their personalities and existing beliefs.

Some people are against abortion for any reason but experience has shown already that many students are either for or against abortion without having given it much thought. The common assumption is that abortions are a matter of convenience only. Many that are opposed to abortion change their opinion when it comes to disorders like Tay-Sachs. They had just never considered a situation like that before. Even those that remain "anti-abortion" become much more accepting of a decision to abort by others after realizing that it is based on a desire to reduce suffering, just as is their own decision.

Because the debates are based on realistic situations in which the students can picture themselves, their willingness to come up with answers (opinions) should be very high. Their readiness, aspiration and attitude levels should be appropriate for the tasks involved in this topic. Anxiety might be a problem unless it is dealt with, which is why I would choose the opinion for the student to discuss, as mentioned earlier.

Thus all five of the previously mentioned motivational factors should cause the students to learn the required material.

Throughout the four topics the factual material and operations built on what went before. So did the ability to apply this material outside the classroom. This transfer of knowledge from the classroom to life is a primary goal of formal education. When it is attained, as it is in this example, there is reason to believe that the willingness of

students to study and to acquire knowledge will increase. Thus greater learning is achieved than would otherwise be possible.

CHAPTER FIVE

EVALUATION

There are two items to be considered in an evaluation; that concerning the students' learning and that concerning the merits of the unit designed to cause that learning. These items overlap, for any benefit given by the unit or course will be partially (but not entirely) reflected in the students' grades. The rest of any benefit will be in attitudes, and this causes some difficulty for it is difficult to evaluate changes in attitudes. By the time a student has been in school 12 to 13 years any attitudes towards school and learning are going to be well established and any changes started by one unit are going to be small and difficult to measure quantitatively.

One can still look for and try to judge qualitative changes in behaviour, and this is what must be done. These are changes that would result from at least partial changes in attitudes due to a greater interest in this unit than in others.

David Pratt has outlined what he calls two "guides" to programme out puts and program aspects, to aid in an evaluation of a curriculum revision. Because I have used only one unit as an example, and most teachers will want to test one or two units by including more applications in the content before committing themselves to revising whole courses this way, I have only included points from his outlines which pertain to unit revisions.

GUIDE TO PROGRAM OUTPUTS AND PROGRAM ASPECTS[60]

I.

Dimensions and Questions	Data Sources
Effectiveness	
What proportion of those registering for, entering and completing the program achieved which objectives, at what level, on completion of the program and in delayed post-testing.	Analysis of program test results; interviews with graduates and dropouts, employers or subsequent teachers.
Acceptability	
Did the students enjoy or appreciate the program?	Analysis of course evaluation questionnaires, discussions with students; disciplinary incidents; attendance data; damage to equipment. Subsequent academic choices of students; subsequent course enrollments; classroom observation.
Efficiency	
Is there any obvious wastage of student or teacher time, of materials or supplies.	In class observations.

II.

Aspects and Questions	
Is the need that the program was designed to meet still significant?	Interviews with learners, teachers, and others in community.
Aims and Objectives	
Are these recognized and accepted by learners? Are they functional, significant and appropriate? Are there additional objectives that could usefully be introduced?	In-class observations; analysis of learning materials and tests; interviews with students.

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[60] D. Pratt, Curriculum Design and Development (Toronto: Harcourt, Brace Jovanovich Inc., 1980), pp. 423 to 425.

Evaluation of Student Learning	
Are performance criteria, formative and summative tests, grading and reporting consistent with specifications in the curriculum? Are they providing necessary information to make decisions about students? Is testing consuming excessive time? Do students regard the tests as fair and useful?	Test analysis; data from guidance services; feedback from parents; discussions with students.
Entry Characteristics	
Are prerequisites and pretests used as intended? Are any students out of place in the program?	Student complaints of difficulty or easiness of program; analysis of test data; student failure and dropout.
Instruction	
Are the content and methods consistent with the curriculum? Are they appropriate to the objectives and the learners? Are they interesting?	Analysis of formative and summative test results; classroom observations; interviews with highest and lowest achievers.
Provision for Aptitude Differences	
Are students' learning problems rapidly recognized, diagnosed and remediated?	As above.
Logistics	
Are materials and facilities that are specified available and being used? Are the materials effective?	Classroom observations; discussions with students.

Many of the data sources are used for more than one purpose. Classroom observations, for example, are used to judge many of the program aspects, and what actually differs is what you are looking at or for during such observations. This guide does provide a plan to follow, as it is intended, but it does not provide any assurance that the "data sources" are reliable.

It is expected that the grades and/or behaviour will be improved over what is normally expected for the genetics unit when the changes I

have suggested are made. This will give support to the idea that content that is more personally applicable to the students will heighten motivation, and in turn lead to greater achievement.

To be honest and accurate in such an evaluation it is necessary to consider some of the faults with the various evaluation techniques.

For interviews, "Although the preparation for an interview is different (from classroom observation), once it is in progress, the method for obtaining information is through observation."[61] Judging of behaviours and attitudes through observations is a very subjective evaluation, particularly when one wants to quantify any changes for comparison purposes. Because the primary purpose of the evaluation is to see if motivation has improved, the observer is looking for improvements in behaviour which may not exist. This means that the evaluator must try to be consistent in his assessments of any course revision. As the judging will most likely be done by the person who has revised the unit he can be tempted or even fooled into seeing what he wants to see.

Facial expressions and tones of voice may allow a secondary appraisal of the honesty of student statements during interviews and discussions but these are not available when one uses questionnaires. For one reason or another, students may not answer these honestly. A virtue of questionnaires is the fact that they allow more time for a fair appraisal of each item than do interviews.

A sample of a suggested questionnaire follows on the next page (Table 5.1). The purpose is to judge whether the students view the topics studied in this unit (i.e. predictions, avoidance of mutations) as useful in their life, or whether they rate their usefulness on the basis of careers.

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[61] Ministry of Education, Evaluation of Student Achievement (1976), p. 29.

TABLE 5.1

STUDENT QUESTIONNAIRE

If the following questions can not be answered with the title of a course or unit, then answer them with a yes or no. Avoid the use of "sometimes".

1. (a) Do you see some courses in school as being of more use to you than others? _____

   (b) If your answer was yes:
   list two that are useful because of your career plans, _____

   _____

   list two that are useful because they apply in your daily life. _____

   _____

2. What kind of a career(s) have you decided on, or are you considering? _____

3. Of all the units taken to date in <u>this course</u>

   (a) which do you consider the easiest? _____

   (b) which do you consider the most difficult? _____

   (c) which do you consider the most interesting? _____

4. Do you consider some units in biology to be of more use in day to day living than others? _____

   (b) if your answer is yes,
   name a useful one _____

   name a non-useful one _____

5. Do you read (or partially read) science books or magazines for pleasure or interest? _____

An alternative form of questionnaire is suggested by Willis and Allen. They required students to select one adjective from a given list of thirty eight. It was to be the "one adjective that most nearly approximated their (the students') reactions to the course for that day".[62] This was also done to describe their feelings for the entire course at various times of recording. The two lists could then be used to pinpoint particularly good and bad lessons or to show changes in the overall view of a unit or course.

Another method of evaluation is the use of a numerical rating scale of the behaviours that one wants to see change. A sample of such a scale is on the following page (Table 5.2). These ratings are of course based on observations, but by limiting the choice of ratings, and by repeating this procedure several times throughout the unit, any changes should become apparent if they occur.

Attendance patterns may show a change throughout a unit or between units and may give a true indication of student appraisals of a unit. Because this is a yes or no type of evaluation it is not open to teacher manipulation, but of course is subject to influence by non-classroom factors (co-incidental illness, play and sport practices) and simultaneous units in other courses.

As can be seen, each of these methods has advantages and disadvantages, and for someone seriously searching for changes in behaviour, no one of them by itself is sufficient.

Evaluation of student achievement will also help to judge the effectiveness of a unit. If the changes I have proposed for the unit do increase

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[62] G. Willis, A.J. Allen, "Patterns of Phenomenological Response to Curricula", Qualitative Evaluation, ed. G. Willis (Berkeley: McCutchan Publishing Corp., 1978), p. 57.

TABLE 5.2

STUDENT BEHAVIOUR

NAME _____

UNIT OF STUDY _____

DATE				AVERAGE
1. Appears happy to come to class	2 1 0	2 1 0	2 1 0	2 1 0
2. Has done or attempted homework	2 1 0	2 1 0	1 1 0	2 1 0
3. Volunteers answers to homework	2 1 0	2 1 0	2 1 0	2 1 0
4. Participates in class discussions (interested enough to have opinions)	2 1 0	2 1 0	2 1 0	2 1 0
5. Asks questions about the topic (wants to understand)	2 1 0	2 1 0	2 1 0	2 1 0
6. Asks questions related to but off topic (wants to know more)	2 1 0	2 1 0	2 1 0	2 1 0
7. Daydreams, talks or appears bored	2 1 0	2 1 0	2 1 0	2 1 0
8. In general, does it appear that the student				
a) seems to like the course	2 1 0	2 1 0	2 1 0	2 1 0
b) seems to like the unit	2 1 0	2 1 0	2 1 0	2 1 0
c) does what is required of him/her	2 1 0	2 1 0	2 1 0	2 1 0
d) does the work because he/she feels they must	2 1 0	2 1 0	2 1 0	2 1 0
he/she wants to	2 1 0	2 1 0	2 1 0	2 1 0

This is to be done for each student at least three times throughout the course of the unit.

motivation of the students as I believe that they will, then corresponding changes in achievement will be observed.

Using achievement as a method of assessing the value of a course or unit change is also not without its faults. Even with no changes to a course, student achievement can vary from year to year for the samples compared in different years are different. Therefore, after a change in a course or unit is made a change in achievement by students in any one year is not a definite indication that the achievement changed because of the "new" unit.

For example, before I made any changes in the way I taught the genetics unit, I had in two consecutive years, medians of student achievement of 78% (with a range of 44 to 100) and 56% (with a range of 11 to 86). This is a case where the content was the same, and because the teacher was the same, the methodology, testing procedures and types of questions were the same and yet there were large differences in the percentages. If such a difference coincided with the introduction of a new unit, it could counteract or exaggerate any changes actually caused by the unit.

A change in the content or approach is not a controlled experiment where only that one variable is changed. The students, with their individual characteristics (goals, abilities, readiness, etc.) differ, and of course outside influences can differ.

Although I believe that adding applications of any material being studied (in this example birth defects and counselling) will heighten motivation and improve both achievement and behaviours, it is essential that anyone making such changes try this for several years before arriving at any conclusion as to its effectiveness.

This repetition of data collection is also necessary when using

the numerical rating scale shown earlier. If it is only used during one (revised) unit, the evaluator might draw the wrong conclusions. For example, some students may ask few questions and/or not complete assigned work, etc. which looks as if the "revision" has not accomplished anything. However, if these same students did not ask any questions before, and never did any assigned work, then it means that the revision has caused some beneficial changes.

A similar comparison should be done before the revisions to a unit or course are carried out, by using the same questionnaire on the un-revised unit. Students generally find some topics of more interest than others, and one could possibly find that even without any revisions, a particular unit induced some significant changes in student behaviour.

For a fair comparison, I suggest that the numerical rating scale should be used in two or three units (e.g. digestion and genetics) before any revision, to compare the differences in student interest in the topic. Then the same units could be rated after the one is revised so as to compare the effects of the revision.

When evaluating the achievement of senior students I do not believe in giving marks for doing homework, for that is all one is doing--giving marks. At this grade level, reinforcement should be from comments, satisfaction, and marks earned on tests and assignments.

All questions on tests and later examinations should correspond to the objectives outlined for that particular unit or topic. During this unit, I suggest a short test part way through the first topic. This would involve preparation and solution of monohybrid type of punnett square and a few basic definitions. The purpose of this test would be to build confidence and raise aspiration levels as much as to measure

achievement.

Longer tests would follow at the ends of topics 1, 2, and 3, and although these are primarily summative in nature, the results should be examined for apparent weaknesses in class knowledge or understanding of particular items. Any such items would then be reviewed so that there is a formative purpose for them.

At the end of topic four (i.e. the ethics portion) the assessment would not be based upon a written test, but rather partly on the debates presented by the students doing part "A" of the questions in that topic. All members of a "team" would get the same mark. The students should be told before-hand that this will be the procedure.

Not only is this more subjective, but in a verbal presentation judging must be done quickly. Therefore, to keep it simple I would use the following plan of marking:

	YES		NO
1. Did the students see all the major implications?	5	2.5	0
2. Did the students appear to weigh the factors in the question?	5	2.5	0
3. Were the answers presented clearly so as to be understandable?	5	2.5	0

The other part of the mark would be for the written answer to question 1 in part B. (See page 78.) The students would be told that one of the answers would be marked, but not which one. The marking of this question would use the same plan as just shown although with more time to study a written answer, partial marks (for example, 3 or 4 etc.

of 5) could be given. Thus the questions in the ethics topic are being treated as an assignment rather than just as homework at practicing a skill.

Because of the time involved in doing their own family pedigree, there is a temptation to give marks for the completion of this as an assignment. Because of the personal nature of the questions and the pontential for embarrassment, the possible variations in the length of it, and the possible variation in the honesty of the students in giving reasons for differing lengths, I do not believe it can be marked fairly. Therefore, I would not use it for evaluating the students. I would consider its completion as a means of evaluating a student's interest in the unit.

The comparisons and tests, I predict, will show that the study of the applications of genetic knowledge and the relevance of those applications to the lives of the students will result in noticeable increase in marks, and a detectable (though temporary for one unit) change in the work habits and interest displayed by the students.

CHAPTER SIX

SUMMARY

The aim of this project has been to show how applications of knowledge can be used by a teacher to increase the willingness factor in motivating students to achieve more learning. Although the example chosen to show this was genetics and related topics, I hope it is clear that I believe the same approach should be used with all science courses and in all years.

Any unit or portion of a course taught with an emphasis on applications will still include most, if not all, the same facts and operations that it would without the mention of applications. By containing applications the unit will therefore have more actual content. Any requirements for the unit to provide general background knowledge or to supply a prerequisite knowledge for future higher education and/or training will still be satisfied.

With the addition of "new" content in the way of applications, the time required for the teaching of this material might appear to be greater. The time can be adjusted by leaving out much of the repetition that now occurs in many courses.

One example of this repetition is the topic of light which is started in grade 9, continued in grade 11, and then completed in grade 13. Each time study of a topic is stopped and then re-started, time must be used to refresh the memories of the students about information they previously learned. Repetition will not be necessary if each topic or

unit is completed in the course where it is started. Moreover, frequently it is only by completing a topic that the uses of that topic can be developed.

Unfortunately, when educators write of applications of knowledge they tend to do so in general or academic terms. For example, what the Ministry of Education considers to be "uses" are usually not seen as uses by the students. As an example, the Intermediate Curriculum Guideline says, in reference to the role of green plants, that students should study, "----their use as a source of food; their release of oxygen to the atmosphere; their use as medicinal herbs;----".[63] This shows that Ministry officials endorse what can be called "uses to the world". This kind of use may explain to the students why society thinks he or she should learn about green plants. However, there is nothing that the student can personally apply to his or her life. In my view, they should also be learning about which plants to grow at home, how to grow them, how to protect them from weather and insects, dangers of herbicides, etc., and possibly about how to do cost comparisons. This is knowledge needed by anyone considering growing plants.

For a variety of reasons, one can expect more differences to exist between individuals in grades lower than the thirteenth. It follows then that the presentation of any one particular "use" of some topic will not be seen by all members of a class as an application for themselves--it just will not be personally applicable to their own life outside of school. Referring back to the last example, it is obvious that all students will not be interested in growing or caring for plants at home.

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[63] Ministry of Education, Curriculum Guideline for the Intermediate Division Science, (1978), p. 150.

This situation does not negate what I have said up until now. It does mean that it will be important to present as many applications as are reasonable for any given topic in order to increase the likelihood of each student finding and accepting at least one of these as applicable to himself. Even if there are still topics from which a student can extract nothing that he can use, if this attempt is made in all units and all courses, it is inevitable that a student will find much that will interest him during his complete science education. Any increase in this awareness is a vast improvement over the situation now, in which students get the right to continue their education, but get little out of their science education in the way of applied or applicable knowledge.

My views find support in Gage. As he says in his chapter on the "Transfer of Learning", "Thus teachers should always try to present a wide array of examples in which the student can see how new principles and techniques can be used. Many of these examples should be placed in real-world settings".[64] Earlier he says, "Do not take it for granted that they [the students] can apply what they learn to new situations. Help them see those applications".[65]

There has been some advancement in this regard in several recent text books. The physical science text by Andrews[66] uses some everyday phenomena to justify belief in the particle theory, although many of the phenomena are still outside the category of student use.

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[64] Gage and Berliner, Educational Psychology, p. 360.

[65] Ibid., p. 355.

[66] W.A. Andrews et al. Physical Science An Introductory Study, (Scarborough: Prentice-Hall, 1978), pp. 94-127.

Birenbaum's[67] and Ashcroft's[68] supplementary texts do try to get across chemistry principles by relating these to real-world settings.

Certainly finding examples of uses of many of the facts and theories that we teach today, particularly when one is concerned with uses to the students, should not be a one man job. No author or teacher can think of as many as groups can. Subject coordinators and consultants should be involving all teachers in this activity, and time should be spent at professional development days organizing and exchanging ideas on applications by the students (in his real world) of the material learned in the classroom. It is also necessary for more authors of texts and particularly those in charge of designing and revising course curricula to do this.

Science is a difficult discipline. In varying degrees, it requires the computational skills and problem solving ability of mathematics, physical coordination for laboratory work, the ability to analyze (and eventually synthesize) ideas, memorization of multiple facts to which references are frequently made, the ability to visualize abstract ideas and patterns, and the ability to both read and write information concerning abstract material.

If we as educators expect to see students putting out the effort and time to actually meet the objectives we set, then we must be more realistic about what we are teaching.

I have tried to show that by relating the content of a course to

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[67] L. Birenbaum, Chemistry of Man and Molecules, (Agincourt: Book Society of Canada, 1975).

[68] K. Ashcroft, Chemistry of Photography, (Agincourt: Book Society of Canada, 1980).

students so that they can see a use for it, they will be more interested in this content. This should by a cyclic process for more interest causes a greater willingness to learn. This willingness in turn will lead to a greater effort and more time expended and should eventually cause more students to have more success at the tasks involved. As the references cited by Simpson[69] show "success breeds success", for it strengthens the students own self image and that in turn causes them to try to do better. This is raising their aspiration levels. With more students liking what they are learning due to its usefulness their attitudes should be one of maintaining their environment. Although anxiety and lack of readiness will still have to be treated on a more individual basis, even these should be partially compensated for within a programme such as I have advocated.

The main factor in starting this cyclic process is student willingness to learn and it is largely based on the usefulness which the student perceives in the subject matter.

It should be realized that this appeal to knowledge that has applications in no way negates or implies any specific method of teaching. Even without curricula changes by the Ministry or Boards of Education, teachers can still change their emphasis and add more to a course so that students can transfer knowledge of principles to knowledge of uses. Teachers who allow students to choose their own content can still do so. Similarly lectures, discussion approaches, or experimental methods of gathering information can still all be used by anyone who prefers a specific method. These methods are all part of the activity portion of

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[69] R.D. Simpson, "Breeding Success in Science", The Science Teacher (November, 1979), pp. 24-26.

the three step learning process and are not themselves the material to be learned.

Teachers should still try to make the activities of learning interesting and varied, but it is the willingness to carry out these activities which is the motivational factor that will determine success.

APPENDIX 1

BEHAVIOURAL OBJECTIVES FOR THE STUDENTS

The appropriate objectives to be accomplished are those following, and are for the most part in the form recommended by Mager[70]. This means that the objective should result in an observable behaviour or action by the student, meet some level of acceptable performance and be accomplished in a specified time.

Specifying the objectives in this manner in no way implies a definite order of presentation or method of presentation.

Neither the times nor the criteria of acceptable performance are specified with each objective for this would be too repetitive. Instead the requirements are summarized here.

Time requirements during evaluation should be based on what seems reasonable for the objective at hand. That is, very short times for writing definitions that should be known; longer times for descriptive answers; and longer times for explanations requiring thought organization as well as knowledge.

When deciding what will be the minimum level of acceptable performance it is necessary to decide first whether part marks or credit will be given for work done by the students.

For definitions there should be no part marks. A definition or meaning is either known or it is not.

For the recall type of objective where a student is to list or

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[70] R.F. Mager, Preparing Instructional Objectives (Belmong, California: Fearon Publishers, 1962).

state four items, for example, if he can only list three, then the mark should only be three quarters of the total mark allocated to that question.

In descriptive, explanatory or decision making questions there will be a number of items expected as indicating the complete fullfilment of the objective. This number will depend on the topic and depth covered in class but any less than this expected number should receive only a fraction of the total possible marks for that question.

Tests given early in the study of a topic may be mainly recall types for the students will not have progressed to the point of using these "facts". But whenever possible, tests should contain some of each type of question listed above. If this is done, it is then up to the teacher or the board to decide what percentage of partially learned objectives will be acceptable. The normal percentage is 50%, but this does not mean that this cannot be different for certain topics.

The objectives listed below are derived from what is stated or implied by the Ministry Guideline (see Appendix 4) and what is needed to understand the topics sufficiently well to apply the knowledge to the world and personal situations.

By the end of each topic, each student is expected to be able to do each of the items listed.

GENETICS:

1. Define (either verbally or in writing) and/or give an example of each of the following words or expressions: dominant and recessive genes, mono-, di-, and trihybrid crosses, hybrid, homozygous, heterozygous, genotype, phenotype, allele, incomplete and co-dominance, carrier, homologous pairs, autosomal and X-linked traits.

2. Explain the following words or expressions: Barr body, mosaic, and deactivated X chromosome.
3. Given the genotypes or phenotypes of the parents and the dominant characteristic, prepare punnett squares to determine the ratio or fraction of possible geno- or phenotypes in the offspring, for mono- and/or dihybrid autosomal crosses, X-linked crosses, or any combination not to exceed a dihybrid cross in size.
4. Given the genotypes or phenotypes of the parents, prepare punnett squares to determine the ratio or fraction of possible geno- or phenotypes in offspring involving traits where co-dominance exists.
5. Given a family history, trace the path of a defect of known type (e.g. X-linked) so as to show who might also have or carry the defect.
6. Given a family history showing affected persons, determine the mode of inheritance of that defect (e.g. autosomal dominant).
7. Predict what trait is dominant from the ratio of offspring traits resulting from simple mono- and dihybrid crosses.
8. State or write (in their own words) the Laws of Independent Assortment and Segregation.
9. Explain the significance of the X and Y chromosomes in determining the sex of humans.
10. State that the normal chromosome number in humans is 46, being comprised of 22 homologous pairs and the sex chromosomes.
11. Describe (to the level covered) karyotype analysis, and chromosome identification by staining.

MUTATIONS:

1. Explain the meaning of the following: mutation, mutagen, carcinogen, teratogen, congenital, gene pool, somatic cell, germ (stem) cell.

2. List the five causes of mutations and/or explain (to the level covered) how each of the causes of mutations (except spontaneous) alters the gene or chromosome structure to produce the mutation.
3. State that spontaneous mutations in humans occur in about 2 out of every 1000 children born.[71]
4. List the four cell types that could be altered, and/or describe the possible outcome from a (noncancerous) mutation in each type.
5. Explain why the size and/or frequency of exposure to a mutagen can effect the risk of a mutation.
6. Explain why most harmful mutations still in existence in the population are recessive rather than dominant.
7. Given a situation where the student would be exposed to a potential mutagen (such as a housing development located on a radioactive land fill site), make a decision as to what he would do, stating the reasons, or state what further information he would need, and why he would want it before making a decision.

GENETIC AILMENTS AND COUNSELLING:

1. State four types of people who should avail themselves of counselling, or given a description of persons, their offspring and/or their family history, identify those who should consider genetic counselling.
2. List the possible types of treatment (other than abortion) that can be used to treat victims of genetic defects, and give at least one example of an ailment where that type of treatment can be used.
3. List at least three minor and three major genetic ailments and state

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[71] J.W. Kimball, Biology (4th ed.), (Don Mills: Addison-Wesley, 1978), p. 598.

what criterion they would use to classify each as minor or major, and show how each meets or does not meet the criterion.

5. Describe the pre- and post-natal tests, including ultrasound and amniocentesis, that can currently be done to diagnose genetic disorders, and/or state any risks involved in each or thought to be involved.
6. Explain why some predictions of risk are hard to determine, by describing how environmental factors can have an effect on gene expression. Accompany this with an example.

ETHICS:

1. Explain the meaning of "genetic screening".
2. List two advantages and two disadvantages of mass genetic screening.
3. Given a situation involving a genetic disorder and a possible course of action, either (a) present arguments both for and against this action, or (b) state and justify what decision the student would make concerning the action.

APPENDIX 2

SUGGESTED TIME SCHEDULE

The following pages show how the various topics comprising this unit can be presented period by period.

By suggesting this schedule it should be understood that this is a guide, and it is not necessary that it be adhered to should a reason for deviation occur.

Discussions may take longer or shorter times than anticipated, questions and explanations may arise unexpectedly. News items that touch on any of these topics may occur. Students may "catch on" quicker than expected. Any or all of these may require a revision of the schedule.

In some cases, changing the order of material presentation may be used to fit shortened or lengthened times. At other times the amount of work done in class and at home can be adjusted to meet the situation.

It should also be understood that this is not a course or unit description. Details about the items listed can be found in either the relevant chapter of this project or in the objectives.

At present, each period is 72 minutes in length.

PERIOD 1

a) Use a recitation method (i.e. combined lecture-question and answer) to explain possible variation in human traits. Give examples.

b) Give a quick overview of the topics involved in this unit and how they will fit together. Emphasize personal and future offspring applications.

c) Introduce punnett squares as an easy method of "book-keeping" when predicting offspring characteristics.

d) Do an example showing the use of a punnett square (more if needed) defining the following words as they arise: homo- and heterozygous, carrier, dominant and recessive, and geno- and phenotype.

e) Students are to do #1 and 2 of Genetics questions.

PERIOD 2

a) Take up questions given yesterday, and review terms.

b) Review distribution of chromosomes and therefore genes as the result of meiosis.

c) Go over rules of probability using series of short leading questions. Summarize these and emphasize the lack of guarantees.

d) Explain the meaning of dihybrid cross and do examples(s) of same.

e) Hand out sheets of birth defect information for the students to use doing questions.

f) Students do #3, 4, 7 and 9 of genetics questions. (See pp. 35-38.)

PERIOD 3

a) Take up questions given yesterday.

b) Give another dihybrid cross question for work in class. Check students as they work and then take this up.

c) Review probability with several questions (verbal).

d) Give and do example of co-dominant situation.

e) Give human chromosome numbers, and explain meanings of alleles, autosomal, mosaic(only if it arises), Barr body, and deactivated-X.

f) Give out PTC Lab for completion at home.

g) Students do questions #5, 6 and 8 of genetics questions.

PERIOD 4

a) Give students a short test involving a monohybrid cross and several definitions.

b) Take up questions given yesterday.

c) Discuss PTC test results. Emphasize the importance and difficulty of determining the dominant trait.

d) Explain karyotype analysis and use of banding techniques for identification.

e) Formally state Mendel's Laws.

f) Give and do example of X-linkage situation.

g) Students do #8, 11 and 12 of genetics questions.

PERIOD 5

a) Return and take up tests given last period.

b) Take up the questions given yesterday, and explain mosaics.

c) Do examples of Pedigree type questions. Refer to PTC lab, and the problems of determining dominant trait.

d) Students do #13, 14 and 15 of genetics questions, and start #1 of Counselling questions. (See pp. 63-69.)

PERIOD 6

a) Take up the 3 genetics questions.

b) Explain mutations, congenital, carcinogens and teratogens.

c) List and explain the different cell types in the body.

d) List the causes of mutations.

e) Group the students and assign the questions from the Mutation topic to various groups. (See pp. 52-54.)

PERIOD 7

a) Test on all of genetics topic.

b) Discuss some of the information from sheets on "birth defects" given out period 2.

c) Students continue to work in groups to complete their answers to the mutation questions. Collect these at the end of the period.

d) Students are to go through the sheets on birth defects and list all possible tests listed and all possible treatments listed for birth defects (by type).

PERIOD 8

a) Take up the possible treatments.

b) Who needs genetic counselling? Discuss and summarize.

c) Check for completeness of their own pedigree (#1 of genetic counselling questions).

d) Return work on mutation questions and let students make additions or changes if needed.

e) If time permits, start the presentation of answers to mutation questions.

PERIOD 9

a) Students from class present their "answers" about the mutation questions.

b) Students are to read the sections in text concerning PKU and sickle cell anemia.

c) Students are to do part C of mutation questions.

PERIOD 10

a) Complete presentations to class.

b) Take up part C and discuss briefly the ideas of gene pool and adaptation.

Sickle cell anemia and malaria should be one of the examples used.

c) Students should leave space to complete mutation topic.

d) Students do #2 and 4 of counselling topic questions.

PERIOD 11

a) Complete mutation topic by discussing sex aberations and Down's syndrome. Give graph of frequency vs. age for the latter.

b) Take up the questions given yesterday.

c) Explain ultrasound very briefly and give out article.

d) Students are to read article and do the accompanying five questions. (See p. 68.)

PERIOD 12

a) Take up the questions concerning ultrasound.

b) Discuss and explain polygenic disorders (e.g. spinal bifida, and hypercholesteremia).

c) Students are to read the article "Who Shall be Born."

d) Students do questions #3 and 5 of the counselling questions.

PERIOD 13

a) Test on mutation topic.

b) Take up the questions given yesterday.

c) Give chart of prediction probabilities.

d) Discuss article and use this to lead into the role of a genetic counsellor.

e) Read article on amniocentesis and do questions 1 to 10. (See pp. 68-69.)

PERIOD 14

a) Go back to previous work assigned and take up the possible detection tests that can be done.

b) Discuss the amniocentesis article and procedure and take up the questions given.

c) Discuss the importance of weighing risks vs. priorities and knowing one's own priorities.

d) Take up mutation test.

e) Describe screening for ailments giving benefits and problems of them.

f) Students are to go through descriptions of birth defects or other material, and list what they consider to be 3 major and 3 minor ailments--with reasons for so judging them.

g) Students are to put down (on paper) their goals in life, and (try to) decide how the birth of a genetically defective baby would alter or interfere with these.

PERIOD 15

a) Take up minor and major ailments. (There is no "correct" answer but there should be reasons for choices.)

b) Discuss goals and possible results of a birth defect. (Keep this from becoming too detailed.)

c) Assign groups for answering part A of Ethics questions and let them work on these. (See p. 78.)

PERIOD 16

a) Give some time for students who worked at home to get together with other group members and get organized for the debates.

b) Start debates.

c) Students are to do part B. (See pp. 78-79.)

PERIOD 17

a) Complete debates if necessary.

b) Take up questions given yesterday, (i.e. part B).

c) Start the next unit.

PERIOD 18

a) Give a test on the defects and counselling and ethics topics.

b) Continue with next unit.

TEACHING AIDS

Films with genetic information are primarily to show DNA structure or meiosis and would most likely be used in those units.

Slides or photographs of victims of birth defects could provide illustrations of symptoms for both observational purposes and as a means of varying some of the content presentation.

One "commercial" source of such slides is the National Foundation of the March of Dimes entitled, "From Generation to Generation: Genetic Counseling". This is available in either a filmstrip (1982 cost of $10 U.S.) or a 124 slide set ($15 U.S.) along with a cassette or record.

Illustrations in texts lack the contrast needed for photocopying even if one were tempted to reproduce them, so they can only be used with an opaque projector. Unfortunately, there is no one text that supplies many suitable photographs unless one is willing to go to a medical book such as the Birth Defects Compendium[72].

Because of this shortage of readily available material, I have avoided any reference to aids such as these. As a teacher finds appropriate material and begins to use it, this use will necessitate a change in the previous schedule.

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[72] D. Bergsma (ed.), Birth Defects Compendium (2nd Ed.), (New York: A.R. Liss Inc., 1979).

APPENDIX 3

DESCRIPTIONS OF SELECTED BIRTH DEFECTS

The following pages show the information that is to be given to the students for doing questions and for extracting information on the severity of a defect and types of tests and treatments that are available.

Information sources are identified by the following code:

[A] = Daniel Bergsma (ed.), Birth Defects Compendium (2nd Ed.), (New York: A.R. Liss Inc., 1979).

[B] = A. Milunsky, Know Your Genes (New York: Avon Books, 1977).

[C] = Huntington Society of Canada, Huntington's Chorea.

[D] = Daniel Bergsma (Ed.), Ethical, Social and Legal Dimensions of Screening for Human Genetic Disease (Miami: Symposia Specialists, 1974).

Numbers following the defect name refer to the defect identification number used in the source quoted.

CYSTIC FYBROSIS (237)[A] autosomal recessive

Frequency: 3 to 5% of general Caucasion population

Risk: about 1 in 2000

Symptoms: Pulmonary diseases (recurrent bronchitis, pneumonia, emphysema).

Blockage of collecting systems in the body (i.e. liver, testicles).

Enzyme deficiencies of the G.I. tract.

Life expectancy is about 12 years for females and 16 years for males.

Tests: Blood test for carriers and/or affected (these do not work as well with heterozygotes as homozygotes).

Sweat electrolyte test by age of 1 month for affected.

Treatment: Pancreatic enzyme replacement
Adequate salt ingestion
Antibiotic therapy (for infections).

CYSTINOSIS (238)[A] autosomal recessive

Frequency: rare Risk: unknown but low

Symptoms: Accumulation of cystine crystals (in bone marrow, lymph, etc.).

Rickets, acidosis, and growth retardation.

Impairment of kidney function.

Death by about 10 years of age.

Tests: At birth: measure cystine content in leukocytes.

Before birth: measure cystine content in cultured amniotic cells (via paper chromatography).

Heterozygotes can be detected but screening is not done.

Treatment: Kidney transplant.

Vitamin K and D doses.

Fluid and alkali replacement.

DIABETES INSIPIDUS (VASOPRESSIN RESISTANT) (287)[A]

Type I. X-linked
Type II. autosomal dominant

Frequency: unknown Risk: unknown

Symptoms: Continual dehydration which impairs mental and physical development.

Tests: Dehydration and urine concentration studies in offspring.

Treatment: Adequate water replacement.

Thiazide diuretics.

If not the vasopressin resistant types, treatment is by inhalation of the hormone.[B]

DOWN'S SYNDROME (TRISOMY 21)[B] chromosomal non-hereditary

Frequency: See age related graph.

There is 8 fold increase if mother has thyroid irregularity.

Symptoms: Nondisjunction of the 21st chromosome results in extra 21st.

There is a variable degree in severity of all symptoms.

Dwarfism and small head.

Mongoloid facial features.

Mental retardation.

Higher frequency (10 to 20 times) of leukemia.

Heart defects.

Early deaths.

Trisomy 13 and 18 also occur but much more rarely. Symptoms are mostly similar.

Tests: Microscopic examination of fetal chromosomes following amniocentesis.

Treatment: Special education, heart surgery, depending on the severity.

DOWN'S SYNDROME (TRANSLOCATION)[B] chromosomal hereditary

Frequency: 2% of all Down's patients.

Symptoms: The same as with trisomy.

Tests: Frequent miscarriages by female provides warning.

Chromosomal banding (staining) and examination following amniocentesis.

Treatment: The same as with trisomy.

GALACTOSEMIA (403)[A] autosomal recessive

Frequency: 1 in every 40 000. Risk: unknown

Symptoms: Cannot digest milk.

Infections, convulsions, gallstones and jaundice.

Cataract development.

Retarded mental and physical development.

Death in a few months if untreated.

Tests: At birth: measure enzyme activity in red blood cells (galactose-1-phosphate uridyl transferase or galactokinase).

Before birth: Carriers have about 50% enzyme activity.

Treatment: Removal of milk and milk products leads to apparently normal development and life.

Cataracts and retardation already present will not be reversed.

HEMOPHILIA[B] — X-linked recessive

Frequency: 1/8600 males
1/10 000 females are carriers

Symptoms: Excessive bleeding due to lack of any one of several blood clotting factors.

Bleeding (internally and externally) usually leads to other complications as well as economic and employment problems.

Tests: Family history.

Blood tests detect 90% of all carriers.

Amniocentesis to detect sex of fetus.

Treatment: Clotting factors are concentrated from donated blood plasma to make cryoprecipitates which are then put into the patient.

HUNTINGTON'S CHOREA[C] — autosomal dominant

Frequency: 1/10 000

Symptoms: There is a great deal of variation in the severity of symptoms.

Jerking of muscles leading to twisting and spasms.

Difficulty in speaking or swallowing.

Mental depression, loss of memory, and carelessness.

Onset is usually after age 30 and before 40.

Death after 12 to 20 years from onset, from complications.

Tests: Family history.

Treatment: Drugs to reduce abnormal movememts.

Drugs to reduce depression and irritability.

HYPERCHOLESTEREMIA (488)[A] — autosomal dominant

Frequency: Relatively high — Risk: 1 to 2 per 1000
1 in 500[B]

Symptoms: Massive cholesterol increases in serum, tendons and tissues.

Premature critical coronary disease for heterozygotes.

Early death for homozygotes.

Tests: Family history.

Test cholesterol levels at early age.

Treatment: By-pass surgery in some circumstances.
Very restricted diet with no animal fats.
Some drugs (i.e. cholestyramine resin[B]).

KLINEFELTER'S SYNDROME (556)[A] Non-disjunction

Frequency: Unknown
Assumed to be 1/1000 in Caucasian population.

Symptoms: Taller than XY males due to continued growth.
Lower body segment is larger than upper.
Complete infertility and impotence.
Average intelligence is reduced somewhat. (1% of all males with IQ less than 90)
Personality and emotional problems.
Reports of neurological abnormalities (i.e. seizures and tremors).

Tests: Abnormally small testes may attract notice.
Prenatally and any time after birth by chromatin positive test of cells.

Treatment: Hormone therapy for hypogonadism (does not reverse effects).
Psychotherapy for emotional complications.

LESCH-NYHAN SYNDROME (588)[A] X-linked recessive

Frequency: Low

Symptoms: Gout and arthritis.
Progressive mental retardation and motor defects.
Aggressive self-mutilation.
Death by 5 without treatment.

Tests: Enzyme assay in cultured cells at birth or from amniotic cells of patient.
Carriers are detectable.

Treatment: Drugs for gout symptoms (i.e. allopurinol).
Restraining of hands and elbows.

MUSCULAR DYSTROPHY (689)[A] X-linked recessive
(childhood psuedohypertrophic)

Frequency: 1 in every 200 000 below age 20.
0 above age 20.

Risk: 1 in every 100 000 (30% appear to be spontaneous).

Symptoms: Degeneration of muscle tissue and its replacement by fatty tissue.
Onset between 3 and 6 years.
Fatal by age of 20 years.

Tests: At birth: measure serum enzymes (creatine phosphakinase).
Family history.

Treatment: None.

Note: There are five other types of M.D., some of which afflict adults. These are not all X-linked.

NEURAL TUBE DEFECTS[B] Multifactorial

Risk: 1 to 2/1000 pregnancies.

a) ANENCEPHALY

Symptoms: Failure of the embryonic neural tube to close at the top.
Abnormal brain growth and development.
Death before birth or within days of birth.

b) SPINAL BIFIDA

Symptoms: Failure of the embryonic neural tube to close at the base.
There is a variation in the severity of symptoms but in most cases there is complete paralysis below the waist.
May be water on the brain.
May be mental retardation.

Tests: Test of maternal blood for alphafetoprotein (AFP) at 16th to 18th week.
Ultrasound to confirm age if previous test is positive.
Amniocentesis if previous test is positive.

Treatment: Surgery to close opening.
Surgery to implant shunt to remove water if present "on the brain".
Physical therapy.

PATENT DUCTUS ARTERIOSUS (800)[A] — multifactorial/environmental (i.e. rubella, prematurity, hypoxia).

Frequency: 1/830 live births.

Symptoms: Connection between aorta and pulmonary artery remains open.
Increasing mortality between 30 and 40 with no treatment.

Tests: At birth: heart murmer or aortogram.

Treatment: Surgery to close opening. Gives normal life.

PHENYLKETONURIA (808)[A] — autosomal recessive

Frequency: 1/15 000

Symptoms: Deficiency of phenylalanine hydroxylase leads to phenylalanine build up.
Mental retardation ranging from moderate to severe.
Sometimes seizures and severe vomiting in infancy.
Shortened life span if untreated.

Tests: Phenylalanine tolerance test (not reliable in 20% of cases).
Blood tests 48 hours after birth.

Treatment: Diet low in phenylalanine starting within the first few weeks of life.
Normal life span with treatment.

POLYDACTYLY (814)[A] — autosomal dominant but with variable penetrance.

Frequency: whites 1/3000
American Negroes 1/200

Symptoms: Have extra digit on hand or foot. Digit can have varying degrees of completeness.

Test: Family history.

Treatment: Surgery.

RED-GREEN COLOURBLINDNESS[B] — x-linked recessive

Frequency: 8% of white males
(less in other races).

Symptoms: No red, green or combinations can be distinguished.

Tests: Family history.

Treatment: None

SICKLE CELL ANEMIA[B] autosomal recessive

Frequency: 1/10 American Negroes are carriers. Risk: 1/400 for A. Negroes.

Symptoms: Hemoglobin becomes increasingly insoluble at low oxygen concentrations and then causes the shape of red blood cells to distort (i.e. sickle shape).

Distorted cells are fragile and rupture.

Increased frequency of jaundice and respiratory distress.

12 to 39% of offspring die at birth.

Tests: Carrier detection tests on blood.

Treatment: Prevention of dehydration.

Prevention of oxygen shortage.

TAY-SACHS DISEASE[B] autosomal recessive

Frequency: 1/30 Ashkenazic Jews are carriers Risk: 1/3600 if Ashkenazic parents.

1/360 000 for others

Symptoms: Lack of hexosaminidase A enzyme.

Normal for first 5 to 6 months and then progressive brain destruction begins.

Loss of motor control.

Eventual blindness and death at 2 to 5 years.

Tests: Blood tests to identify carriers.

Prenatal diagnosis of fetus.

Treatment: None.

TUBEROUS SCLEROSIS (975)[A] autosomal dominant but with variable penetrance

Frequency: 1/100 000 Risk: about 85% are new mutations.

Symptoms: External nodules.

Internal nodules causing possible kidney and lung impairment.
Calcification of basal ganglia in brain.
Epilepsy and moderate mental retardation in 75% of cases.
Possible death by about 25 years.

Tests: Family history.
X-rays of skull and brain for deposits.

Treatment: Anticonvulsant drugs.
Some surgery.

TURNER'S SYNDROME (977)[A] Non-disjunction

Frequency: 1/10 000 live females
1/500 conceived.

Symptoms: Short stature.
Shield-like chest and webbed neck.
Sterility and sexual infantilism.
Hypertension in 27% of patients.
Cardiac and/or vascular lesions in 15%.
Mental retardation in 36%.

Tests: Chromatin tests after birth.

Treatment: Hormone therapy from second decade until usual age of menopause.
Counselling for psychological problems.

XERODERMA PIGMENTOSA (1005)[A] autosomal recessive

Frequency: Low.

Symptoms: Photosensitivity and photophobia.
Inability to repair DNA damage after ultra violet exposure (from sun).
Usually death from cancer or infections by age 20.

Tests: None.

Treatment: Avoidance of sunlight.
Some surgery.

APPENDIX 4

The following pages are copied from the course of study for Grade 13 Biology (1969) from the Ontario Department of Education to show the relevant topics of the biology course as they are currently described.

Unit I

Characteristics of Living Things

This unit is intended to organize the previously acquired knowledge of plants and animals to focus attention on those characteristics which make them living things.

1. Introduction

Examination of specimens and discussion from which students may prepare a list of the characteristics which distinguish living things from dead, dormant, and fossilized organisms and from inorganic matter.

2. Ways in Which Organisms Move

Observation of at least one living specimen to illustrate each of the following types of movement.

- Muscular movement with skeletal support: vertebrate, arthropod
- Muscular movement with no skeletal support: earthworm, hydra
- Ciliate and flagellate movement: paramecium, chlamydomonas, bacteria
- Distinction between locomotion (above) and motion as in eyelid, eye, tongue, heart
- Movements of higher plants: closing of leaves and flowers, growth movements

3. Ways in Which Organisms Respond to Stimuli

Observations of some of the responses of a living animal and a sensitive plant:

- Nature of stimuli: light, moisture, gravity, heat, cold, sound, touch, chemical substances
- Irritability of organisms without special sense organs
- Special receptors for stimuli: e.g., eye, ear, temperature receptors, pressure receptors, taste organs
- Mechanisms of conduction and response

4. Ways in Which Organisms Reproduce

Capacity for reproducing their kind:

- Asexual methods: vegetative methods or fission to include discussion of one of amoeba or hydra, one of plant bulbs, cuttings, bacteria or chlamydomonas, and one example of spore formation (mushrooms, mosses, or ferns)
- Sexual methods: fusion of gametes to produce a zygote from which the organism develops by growth, division, and differentiation of cells in both plants and animals

5. Ways in Which Organisms Maintain Themselves

A brief discussion of metabolism, its phases (anabolism and catabolism), and its processes: photosynthesis, digestion (extracellular, intracellular), assimilation, respiration, excretion.

Unit II
Cells

This unit emphasizes that the basic unit of life is the cell and that basic energy changes must proceed within it. The living protoplasm in all cells has similar physical and chemical properties. All cells are produced from pre-existing cells.

1. Structure of Cells

- Student use of the microscope to see and review details of cell structure
- Examination of electron photomicrographs to see details of ultrastructure of cells
- Discussion of structure and functions of: nucleus, nucleoli, mitochondria, ribosomes, lysosomes, endoplasmic reticulum, Golgi body, ground substance, membranes, cell wall and pits, centrioles, vacuoles, plastids, starch bodies, fat bodies, protein bodies

2. Composition of Protoplasm

- Review of the structure of atoms of elements important in biology
- Molecules and chemical bonds, ionization
- Chemical reactions and the energy changes associated with them, equilibrium
- Enzymes as biological catalysts: laboratory exercise to illustrate organic molecules; functional groups important in organic reactions: hydroxyl, carboxyl, aldehyde, and amino
- Formation of carbohydrates, fats, and proteins: stress the general structure of the molecules indicating important linkages: ester, glucoside, and peptide. (Formulae should only be used where they illustrate linkages significant in biology.)
- Nucleic acids and nucleoproteins: brief reference to nucleotide bases and their linkage

3. Physical Properties of Protoplasm

- Include colloids, sol-gel changes
- Laboratory exercise to show coacervates and protoplasmic streaming

4. Catabolism

(a) Digestion: laboratory exercise to illustrate the hydrolysis of carbohydrates, fats, and proteins to produce simpler soluble substances (treatment as an essential preliminary to respiration)

(b) Respiration: the oxidation of the glucose molecule to carbon dioxide and water with energy made available in the reactions, laboratory exercises to illustrate

- Glycolysis: phosphorylation, splitting of the 6-carbon molecule to form pyruvate, a 3-carbon frag-

ment; formation of acetyl-coenzyme A; Krebs cycle for controlled release of energy by step-wise breakdown of molecules

- Transfer of energy by the ADP-ATP system, efficiency
- Metabolic role of vitamins and minerals
- Reference to the fact that after preliminary processing into 2-carbon fragments, fats and proteins may enter the same metabolic pathway as glucose
- Role of mitochondria

5. Anabolism

(a) Photosynthesis: laboratory exercise to show the relation of energy and chlorophyll to starch production, involvement of gases

- Energized chlorophyll: transfer of energy to the ADP-ATP system
- Light and dark reactions, formation of organic compounds
- The concept of available potential energy in the molecules
- Role of chloroplasts

(b) Other syntheses: brief treatment to show how simple organic compounds react to form cellulose and glycogen, how they can be converted into fatty acids and glycerol for fat synthesis, and how amino acids can be formed for protein synthesis.

6. Functions of Cell Membranes

Osmosis: laboratory exercise, cell turgor, permeability to substances other than water

- Active transport and its dependence on metabolic energy
- Importances of cell environment, composition of environment, limits of tolerance to changes in medium of cells, osmotic value, wilting
- Laboratory exercise to illustrate plasmolysis

7. Mitosis

- Laboratory exercise to study chromosome changes in growing cells
- Explanation of action of chromosomes and genes
- Self-replication of DNA and its relation to mitosis
- Regulation of metabolism, growth, and development
- Mechanism of virus action

Unit III

Organisms

In this unit the student becomes acquainted with the structures of a highly developed plant and an animal, both consisting of complex systems made up of organs, tissues, and cells. Emphasis is placed on the harmonious interaction of all the systems in maintaining homeostasis for the benefit of the entire organism. Similarities in essential activities are seen in plant and animal.

1. Organization of Differentiation of Cells

Meanings and examples of tissues, organs, and systems; problems of multicellular organization.

2. Dissection and Study of a Mammal

An examination of organ systems and some of the tissues composing them (epithelial muscle, connective, nerve, glandular); a discussion of the gross functions of these systems and tissues with attention to the fact that certain systems function to serve all cells of the body; integration of body functions

(a) Digestive system: mouth, esophagus, stomach, small and large intestines, digestive glands (names of enzymes and their specific actions not essential)

- Function: intake of food, extracellular digestion of food to components that can be used by cells, transfer of end products to blood and lymph for distribution

(b) Circulatory system: heart, arteries, arterioles, capillaries, venules, veins. Note that it is a closed system, that blood circulates, that arterial blood pressure is maintained, that no cell is very far from capillaries of this system (use living frog or fish to demonstrate).

- Block diagram of pulmonary and systemic circulation (names of arteries and veins not essential)
- Lymphatic system
- Function: transport of needed materials, including secretions of endocrine glands, to cells and of wastes away from cells

(c) Respiratory system: nasal passages, larynx, trachea, bronchi, bronchioles, lungs, alveoli, chest cavity, diaphragm, rib muscles, respiratory centre, and main respiratory nerves

- Function: ventilation of lungs, bringing fresh air close to capillaries, where oxygen enters blood for distribution to all cells and carbon dioxide carried from cells leaves blood.

(d) Elimination systems: kidney, glomerulus and capsule, tubules, ureter, bladder, urethra

- Function: filtration of blood, tubule reabsorption of water and useful solutes, elimination of waste solutes, regulation of water and solute content of body fluid which bathes body cells

- Elimination functions of sweat glands, respiratory system, digestive system; brief mention of function of sweat glands in regulation of body temperature and salt balance.

(e) Muscular and skeletal system: muscles, bones (names and descriptions of individual muscles and bones are not essential). Tissues: muscle and connective tissue, bone and cartilage

- Functions: movement, support, protection

(f) Reproductive system

- Male: structures, tissues (gonadal, glandular, muscular), functions
- Female: structures, tissues (gonadal, glandular, muscular, uterine epithelium), functions

(g) Co-ordinating systems

- Nervous system: brain (main parts), spinal cord, motor nerves, sensory nerves, mixed nerves, sense organs (details of structure of sense organs not essential except for the eye). Prepared dissections, rather than class dissections of the brain and spinal cord, could be used.
- Neurons: motor, sensory, connector; structure (cell body, axon, dendrite); reflex arc; associated connective tissue. Function: receiving information from environment and from all organs of the body and co-ordinating rapid response to conditions in environment and internal organs
- Student dissection of a mammalian eye and a study of the functions of its parts; relation of the optic nerve to the visual area of brain
- Endocrine system: complexly interrelated system of glands producing special substances (hormones) which are liberated into the blood and act on target organs. Hormones co-ordinate processes of metabolism, growth, and reproduction and regulate activity of organs which control water and salt content of body fluids. Names and locations of the endocrine glands, the hormones produced, their target organs and their functions
- Relation of nervous and endocrine systems
- Steady-state control of the whole organism: chemical control of breathing rate and depth; control of water balance, salt content, and glucose concentration of body fluids; control of the menstrual cycle in the human female. These control systems should be treated in sufficient detail that general principles of feedback systems will be made clear.

3. Dissection and Microscopic Examination of a Herbaceous Dicotyledonous Plant

Discussion of the following systems and their gross functions, showing that each system serves all the cells of the plant body; disucussion of other dicotyledonous plants to show features not illustrated by the example selected

(a) Supporting system: temporary (collenchyma tissue),

permanent (sclerenchyma tissue, secondary wood), turgor, cell wall

(b) Anchoring system: roots, underground stems, diffuse or tap form of system, advantages of each, extent of root hair growth

(c) Absorbing system: root hairs and osmosis

- Water: water loss and osmotic values (transpiration)
- Ions: Accumulation and rejection of ions

(d) Conducting system: no cell in the plant is far from conducting cells

- Water and minerals: path from root hair to xylem of root, stem, leaves
- Sugars and amino acids: from site of photosynthesis through phloem of leaves, stem, roots, flower and fruit

(e) Growth system: nature and location of the primary meristems and their functions during elongation and differentiation

- Secondary meristems (vascular and cork cambiums) in roots and stems

(f) Gaseous exchange system: stomates and their operation, intercellular air spaces, wet cell walls, gaseous diffusion contrasted with breathing

(g) Photosynthetic system: favourable position of chlorophyll-bearing cells, each cell adjacent to an air space

(h) Food storage system: cells and structures specialized for storing energy in organic compounds: starch, fats, oils

(i) Reproductive system: flower, carpel, ovule, egg, anther, pollen grain, sperm, pollination, fertilization, seed, fruit

(j) Co-ordinating system: auxins operating through growth regions in roots and stems

- Abnormal growth due to disease organisms and parasites
- Initiation of flowering stage triggered by photoperiod and causing changes in the metabolism and growth pattern of the plant
- Effect of environmental factors on the seed development, germination, and growth

(k) Excretory system: waste accumulation in leaf, leaf fall, toxic substances exuded from roots

(l) Regulation of internal environment: plant starch-sugar balance and osmotic effects, water and salt balance, control of pH and temperature

Unit IV

Classification of Organisms

This unit provides a brief introduction to the purpose and principles of classification of organisms.

(a) Brief history of systematics: Linnaean system, present system

(b) Discussion of the principles involved in classification of organisms; meaning of species, genus, family, order, class, phylum, kingdom, as illustrated by the classification of a common example like the domestic cat

(c) Laboratory exercise: use of a simple dichotomous key to identify common Ontario conifers or other species of interest, plant or animal

Unit V

Interdependence of Organisms

This unit deals with organisms as mechanisms driven by energy which is derived from the sun and often stored in energy-rich foods by green plants. As well as energy, other interrelations lead to an appreciation of interdependence of organisms and their relation to the physical environment.

(a) Photosynthetic activity of chlorophyll-bearing plants producing energy-rich foods, carbohydrates, fats, proteins

- Dependence of fungi, bacteria, herbivores, carnivores, on energy stored in this food supply

(b) Food chains and food webs

- Carbon cycle, nitrogen cycle, cyclical use of other elements
- One-way flow of useful energy, pyramids of number, biomass energy

(c) Communities of organisms

- Succession in communities
- Interaction of organisms and environment

(d) Overproduction of offspring; number of spores, eggs, seeds, larvae or nymphs, juveniles, compared with number of adults in populations of insects, fish, trees, etc.

- Fluctuations in population density, natality versus mortality
- Biotic potential versus actual population

(e) Discussion of closely interdependent organisms such

as parasites, saprophytes, symbionts, commensals, and social organisms, stressing the effects on each organism from the association; one example to illustrate each.

(f) Man as a dominant species, his dependence on other species, his effect on his environment, finite sources of energy, infinite potential to reproduce, problem of population size.

Unit VI

Heredity and Variation: Genetic Continuity

This unit describes the fundamental process providing the mechanism for continuance of life of each species by providing new individuals. Attention is to be focussed on the mechanism allowing for variations within populations and the importance of this for heredity and evolution.

(a) Duplication of an individual organism by asexual or sexual reproduction; asexual reproduction including the production of individuals whose inherited material (DNA) comes from a single parent with the chromosome number unchanged (vegetative reproduction) or with a reduction in the number of chromosomes by meiosis. In sexual reproduction the new individual develops from a zygote produced from the fusion of inherited material (DNA) contained in the gametes from two parents; sexual differentiation of gametes in many organisms

(b) Meiosis, variation in allele composition in gametes (e.g., egg and sperm); syngamy (fertilization), zygote, significance of these processes in transmission of genes (DNA) from generation to generation

- Mendelian inheritance
- Alternation of generations treated generally to show the occurrence of the haploid-diploid cycle in plants and animals, reference to the existence of tetraploids and polyploids
- Sex chromosomes, sex determined by a specific group of genes

(c) A general discussion of the start of life for an individual organism, cell enlargement producing growth, genetic control of these processes, importance of cell environment

- Spore, seed, and egg size in terms of stored energy for young organisms
- General discussion of the differences between plant and animal differentiation and growth, limited in plants to the development of the embryo in the seed, in ani-

mals to the development of the three primary germ layers of the gastrula

- Relative importance of sexual and asexual reproduction in maintaining populations of plants and animals, relation of numbers of offspring to parental care in maintaining population

Unit VII

Evolution: Changes in Living Things Through Time

This unit involves a study of the effects of heredity and of environmental changes on plants and animals.

(a) Darwin's theory of evolution based on evidence of fossils, plant and animal geography, and comparative gross morphology

(b) Variations in population of a species due to non-heritable environmental effects and due to heritable variation

- Recent ideas of mutation (change in DNA) as the source of change in heredity
- Selection of heritable variations due to environment and population pressure as a cause of evolution
- Conservative and divergent action of selection
- Genetic isolation and origin of species

(c) Source and development of domestic plants and animals, human selection in plant and animal breeding

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