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Arno G. Motulsky, M.D.
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Dear Arno:

I really am sorry not to be able to be at the meeting of your committee. I hope it will be a good meeting and that something will come of it all. I agree with you that the American Society for Human Genetics can perform an important function in this way.

It seems to me that you have listed most of the things that will be important to talk about. In what follows, I'd like to make a few comments about these and one or two other things. I'll take them up more or less in the order in which they appear in your list.

I hope that screening for genetic defects will continue to thrive and that new methods will become available rapidly. The therapeutic implications of early discovery of metabolic disease need no discussion. As you have mentioned, however, it is important to press against legislation requiring expensive screening methods for diseases that are very rare. On the other hand, legislative bodies can do something about screening through providing money for improving existing methods and investigating the possibility of new ones.

My own view on genetic counseling is that it ought to be woven into the natural and normal activities of the physician. The interposition of a geneticist between the doctor and his patient I think is wrong. What is required, therefore, is to teach all medical people enough genetics so that genetic counseling is as natural as counseling about anything else. So the society might press in various directions for educational improvement. A couple of years ago, I looked through 64 catalogues of American Medical Schools which I found in the Johns Hopkins Medical Library. I found that

only 4 listed departments of genetics and that of the rest a required course in genetics was listed for only 17, while among those not requiring such a course, electives were listed by an additional 15. These electives were commonly given in departments of Anatomy, Medicine, or Pediatrics. This is not to say that genetics is totally ignored by those medical schools which did not list any courses since obviously departments of microbiology and biochemistry and so on do concern themselves with some aspects, but it really does suggest a lack of recognition, even as late as 1966, of the relevance of genetic principles to medical training. It might be said that genetics is not needed in medical schools since so many students have such good courses in college and in high school and no doubt there is some truth in that although not as much as some might think. The fact is that many people come to our medical school without any genetics at all. This creates a problem for us in giving our course which we meet by spending the first couple of weeks with only those who have not had any genetics and going over the fundamental principles. It seems to me that the society might press the Association of the American Medical Colleges to require genetics for admission to medical school and to do something about teaching genetics relevant to human material in the medical school.

Empiric recurrence risk registers would I think be meaningless. Such risk figures are little better than telling a parent that the probability of repetition of the event is greater than if it had never occurred. The data are usually taken from several sources in the literature without regard for possible differences in collection or even of incidence of the disorder in that part of the world. One never really knows whether the risk figure applies to a particular family.

As for genetic indications for abortion and fetal diagnosis, I think a lot can be done here. Whatever can should be done to support the development of methods for cultivation of amniotic cells and for assays of enzyme deficiencies or other disorders on these cells. This ought to be pushed vigorously because it would mean that it might become possible for families who have had one child affected with some disease or other to have any number of additional children who would be unaffected since affected fetuses would have been aborted.

You might also want to discuss something under a sort of general heading of genetics and the law. That is, as we discover more about the effect of the constitution on personality development the legal profession may express increasing interest. The question is, is a man who has 2 or more Y chromosomes really responsible for his actions, or is he driven by his genes to antisocial behavior? I'm sure we don't know the answer to that. No one, so far as I know, has studied the cultural background of these individuals to see whether it differed in any material way from those of other antisocial persons. But whether it does or doesn't it's quite possible that lawyers will begin to look carefully at the genetic background of persons that they intend to defend or to prosecute. Dr. John Money has discovered that people with XO chromosomal constitution have difficulty with direction sense and with numbers. Doubtless, these perceptual defects could have an important bearing on the personality

structure of such individuals. Reading disabilities and learning defects, appear also to be genetically influenced, if not single gene entities, and it takes no particular imagination to conceive that we will be learning a great deal about the genetics of personality behavior, learning capabilities and so on before very long. Whether or not a person will be able to claim immunity from the law because of his genes is a question that may not come up very often but it might be something that the society could think about.

As for the future, I think the topics you have given are ones which are certainly being widely discussed. I like Muller's ideas least of all because I think breeding of human beings like animals can easily lead to troubles as it has done with animals. I have been looking up the papers on porphyria in cattle recently. Most or all of the cases can be traced to a single bull. Admittedly there is a good deal more inbreeding of cattle than with human beings but the dissemination of sperm originating in single individuals to many recipients might have the same effect. The porphyria gene spread through a good many herds and produced a good many animals which had to be destroyed. Obviously, all of these animals were being bred for desirable characteristics just as the human beings would be bred for desirable characteristics.

To my mind, of all of these things, the most important is education. I would agree that we have made progress and that many medical people now know what chromosomes are and what genes are and what they do. But there is still a general misconception that to study patients who have metabolic disorders, which by chance are caused by genes, is all that one needs to know about genetics, and the larger issue of the causes of human variability of all kinds is given less attention. Medicine is moving rapidly into an increasing concern with the preservation of health. Hopefully it will move beyond that into the means for exploitation of genetic potentialities. Somehow it seems to me we are spending more time talking about genetic surgery for rare diseases than we are about gathering the information to discover genetic potentialities so that we can design appropriate environments for normal individuals. As much concern should be expressed about the latter as about the former, probably even more. But on this issue medical schools are silent.

Again, I am sorry I won't be there. I wish I could be. With best regards,

Yours very truly,

Barton

Barton Childs, M.D.

BC/nch