YOU, YOUR CHILD, AND MENTAL HEALTH

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YOU, YOUR CHILD AND

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PREFACE

An attempt was made to gather information that would be of interest to expectant parents concerned with the mental health of their unborn child. Probabilities of different genetically induced mental diseases were given, causes of genetic mental diseases, how mental diseases can be detected, and the future role of genetics in eradicating mental disease.

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CHAPTER I

INTRODUCTION

The major concern of an expectant mother is not the sex, but the health--particularly mental health--of the unborn child. Questions she may ask herself are, "What causes a child to be born mentally retarded, or to develop mental illness in later life? What are the chances my baby will be mentally healthy? What can I do to insure good mental health for my child?" Scientific studies, together with technological improvements and medical research, can now help provide answers to such questions.

The point of morality aside, parents now have the option of terminating unwanted pregnancies. Because of the availability of abortion, many parents are becoming actively interested in determining the health of a child--particularly mental health--during the prenatal stages. Information concerning how mental retardation and mental illness are inherited, the incidence rate of inherited mental retardation and mental illness, and other related genetic inbalances which may cause poor mental health could be helpful to expectant parents.

An attempt has been made to gather information concerning heredity and mental health and put it into a form that

would be useful to the average parent concerned with the mental health of an unborn child, or with the probabilities of a child developing mental illness in later life.

While great strides have been made in determining causes of certain inborn mental retardation less success has been had with determining the causes of mental illnesses which may develop at anytime during an individual's life time. But, while there are many moral and legal aspects to consider, the future for the study of genetically controlled mental retardation and mental illness has great potential. A consciousness of the basic principles of heredity should be a prelude to the presentation of the role of genetics in mental health.

CHAPTER II

A REVIEW OF THE BASIC PRINCIPLES OF HEREDITY

Genetic influences are transmitted to the child by elements within the parents' sex cells. Chromosomes, microscopic rod-shaped protein and DNA bodies within the sex cell nucleus, are the primary mode of hereditary transmission. Genes, which carry the coded genetic information that will determine the mental, physical, and constitutional characteristics of the new individual, are arranged in a specific order on the chromosomes. It is the genes which determine the outward appearances--such as hair color, height, body shape, as well as the internal mental and metabolic processes.

In humans there are 46 chromosomes in each normal cell of the body except the sex cells, which have half that number, 23. The set of 46 chromosomes consist of 22 matched pairs (autosomes), and one pair of sex chromosomes. In the male the sex chromosome pair consist of an X chromosome and a smaller Y chromosome. In the female the sex chromosome pair consist of two X chromosomes. One of each pair, 23 chromosomes, is inherited from each of the individual parents.[1,2]

As stated, genes contain the genetic information for inherited mental, physical, and metabolic characteristics. These genes occupy certain positions on the chromosomes; thus, as chromosomes are matched in pairs, genes will be also. Although the genes are matched in position for characteristic, they may not be identical for characteristic. For example, the gene for eye color is located at a specific site on the chromosome inherited from the father. On that chromosome's mate, inherited from the mother, the gene for eye color is located at the same site, but the gene for eye color from the father may be for brown eyes and the eye color from the mother, blue. When genes are not identical for characteristic, one may mask or hide the characteristic of the other. That gene characteristic is said to be dominant, and will be manifested in the offspring. The characteristic that is masked or hid is said to be recessive, and in order for a recessive characteristic to appear in the offspring, the genes must be identical. Both parents may carry a recessive gene for a trait which would not be evident, but should the offspring inherit the recessive gene from each parent the trait would become obvious.[1,2]

All 22 pairs of autosomes and the sex chromosome pair have been identified and labeled, and the human gene count is estimated somewhere between 50,000 to 100,000 genes. In the preface of the fifth edition of "The Metabolic Basis of Inherited Disease" it is stated that as early as 1982, the chromosomal location of only 350 genes had been identified.

All of the 22 autosomes, as well as the X and Y chromosomes, contain at least one known gene.

Each of us began as a single cell. A sex cell, with 22 autosomes and one sex chromosome, from the male fertilizes a sex cell, with 22 autosomes and one sex chromosome, from the female and forms a single cell, the zygote. The zygote is no longer a sex cell, but is now a somatic cell with 46 chromosomes--22 pairs of autosomes and one pair of sex chromosomes. The somatic cell, through a process called mitosis, duplicates its chromosomes and divides into two identical daughter cells. Each of these cells will duplicate and divide similarly: one cell becomes two; two, four; four, eight. . . . Gradually various cells differentiate from the mass and become distinguishable as appendages, organs, and physical features. Most of these somatic cells will continue to duplicate and divide throughout the lifetime of the organism, promoting growth and providing new cells as old ones wear out.

When the reproductive organs are formed in the fetus cells will differentiate to become the sex cells. These cells will divide by a process called meiosis. During meiosis the 46 chromosomes of the immature sex cell arrange themselves in 23 pairs at the center of the cell nucleus. At a specific point in the meiotic process, the two partner chromosomes separate, with only one of each pair going into the two resulting daughter cells. Thus, meiosis produces sex cells with a reduction in the number of chromosomes,

from 46 to 23.

The primary formation of sex cells in the reproductive organs of females occurs in the prenatal stage. At this time all the sex cells to be released during the monthly cycle of a woman's reproductive years are present. The female sex cells, the ova, contain 22 autosomes and an X chromosome.

The formation of the sex cells (sperm) in males occurs at puberty and continues throughout the lifetime. The male sex cells contain 22 autosomes and either an X chromosome or a Y chromosome.(1,2)

When the sperm with 22 autosomes and one sex chromosome fertilizes the egg with 22 autosomes and an X chromosome, the zygote again has the full complement of 46 chromosomes, and a new generation has begun.

How, then, can two mentally healthy individuals parent a mentally retarded child or a child who will develop mental illness in later life? Causes of mental disorders by chromosomal abnormalities, gene mutations and environmental factors will be considered in the next chapter.

CHAPTER III

GENETIC CAUSES FOR MENTAL RETARDATION

Genetic causes of mental retardation generally fall into one of three catagories: chromosomal disorders, single or multiple mutant gene action, and outside or environmental factors. Seventy-five percent of the genetic type of mental retardation are due to chromosomal disorders, about twenty percent are caused by mutant gene action, and five percent by environmental factors.(3)

An excess of chromosomes will occur when one pair of chromosomes fails to disjunction or to separate properly during meiosis, so that instead of one of the pair ending up in each of the two resulting daughter cells, one daughter cell will have both chromosomes and the other will have none of that pair.

The most frequently encountered chromosomal abnormalities occurring among live-born infants is the Down Syndrome. Ninety-two percent of children showing Down Syndrome symptoms have been found to possess an extra autosome, chromosome 21. The incidence rate occurring among live-born infants is calculated to be about 1/600, but calculations based on studies of spontaneous abortion suggests the incidence at conception may be about 1 in 200. The degree of

mental retardation of Down Syndrome ranges from moderate to severe.(4)

Edwards and Patau Syndromes are also attributed to an autosomal excess. Children with these syndromes are severely retarded, and experience early infancy death. The incidence rate for Edwards Syndrome is 1/3000 births, and for Patau, 1/5000.(3)

When the sex chromosomes fail in disjunction during meiosis the offspring may experience mental retardation of varying degrees. Males with an extra X chromosome, XXY, are classified as Klinefelter Syndrome, and often show below normal intelligence. Incidence rate for Klinefelter Syndrome is 1/450 males. Studies by J. Phillip Welch indicate that males with XYY syndrome may not be mentally retarded but do show a predisposition towards "antisocial" behavior. Incidence rate for the XYY syndrome is 1/1000 males. In females there is evidence that the Triple-X abnormality predisposes to significant mental retardation and the incidence rate is 1/1000 females.(5)

An individual lacking a sex chromosome, XO, is classified as having Turner Syndrome. The incidence rate is 4/10,000 and the XO individual shows no greater predisposition for mental disorders than the general population.(4)

The exact cause of too much or too little chromatin material is not known. However, studies show there is a noted relationship between maternal age and the frequency of chromosomal abnormalities. Although women over 40 produce

only 4 percent of all babies, 40 percent of all mothers who bear children with chromosomal disorders are over 40 years of age.(4)

But age is not the only factor. Younger women have given birth to Down Syndrome children but in lower frequency than the older aged women. Also, there appears to be no relationship between maternal age and the number of infants born with a missing chromosome.

Chromosomal abnormalities do not appear to run in families, but there are always exceptions. According to genetic studies a sibling of a person with Down Syndrome would be no more prone to have a child with the disorder than anyone else in the general population: the frequency of incidence being 1 in 600 newborn infants.(4)

The basic chemical structure of the chromosomes appears to be remarkably stable, but sometimes a gene, for no apparent reason, will mutate or undergo change.

Usually the effects of mutation are destructive, and the condition arising is often self-limiting due to its severity and the inability of the affected individual to reproduce. However, some harmful mutations can be transmitted through generations, for example by the symptomless carriers of recessive genes. It is supposed that all of us carry several very harmful but 'silent' mutations, hence it is not necessary for each affected person to have an affected parent.

When a mutation occurs in the sex cells the mutant gene does not affect the person in whom the mutations occurs, but

becomes apparent in subsequent generations. If the mutation occurs in the somatic cells at an early developmental stage, it affects the person having the mutation, but is not passed on to the next generations.

About 20 percent of genetic types of mental retardation are caused by gene mutations. Gene disorders may result in metabolic errors which will cause mental retardation. When the metabolism is altered the chemical and physical processes of the body are unable to function properly.(3)

It is believed that all biochemical processes are under genetic control, and these biochemical processes are broken down into a series of individual stepwise reactions. Each biochemical reaction is under the ultimate control of a different single gene, and if that gene should mutate there is a change in the ability of the cell to carry out that chemical reaction.

When the biochemical processes are interrupted the body's ability to properly utilize carbohydrates, proteins, and fats is altered, and a harmful build-up, or a lack of essential substances in bodily organs may result.(6)

Currently there are about 1350 single-gene determined diseases, many of them associated with mental disorders. Phenylketonuria (PKU) is one of the most common forms of metabolic error causing mental retardation. The relationship between Phenylketonuria and mental retardation was discovered in 1934, and since then other mental diseases have been associated with faulty biochemical processes such

as enzyme defects, glutamine deficiency, utilization of mucopoly-saccharides and mucolipids, and the transport of substances to target organs. PKU has been traced to a single, recessive, autosomal gene which regulates the body's ability to metabolize certain amino acids, and in the United States it is estimated 1 of every 50 individuals carries the recessive gene for PKU.(4)

A PKU infant may develop normally the first few months of life and then become dull and lethargic. Untreated PKU will cause severe mental retardation, but with proper diet the symptoms of agitated behavior, irritability, and uncontrolled temper may be decreased or prevented. However, once mental impairment has occurred, it is irreversible. That is why most states now have complusory screening of newborns, and it should be noted--it is the defective gene that is inherited, not the disease.

Environment plays an important role in good mental health also. Although the potential of what a child will be is inherited, there can be no genetic expression without environment. From the moment of conception, heredity and environment are always interacting with one another--always influencing one another.

Possibly, the intrauterine environment is the most important place for controlling mental health. In her book, "At Highest Risk," Christopher Norwood vividly brings to mind the physical damage cigarettes, alcohol, aspirin, and chemical inhalation or ingestion can do to a developing fetus. More

recently it has been learned that such things as levels of loud noises and high stress can influence fetal physical development.

It is suggested that factors such as high stress levels, drugs, chemicals inhaled or ingested, and loud noise levels during early fetal development could alter the genes in such minute ways there would be no noticeable difference in the child, but at some point in life environmental pressures would overload the circuit, causing a breakdown in biochemical processes and resulting in mental disorders.(7)

While we need not be hasty about resuscitating old wives' tales about "marking" the baby, there might be more to the advice than meets the naked eye.

CHAPTER IV

MENTAL ILLNESS

Mental retardation refers to subaverage general intelligence and mental illness has to do with the way people behave when actions are based on emotions and irrational ideas instead of reality. Genetic mental retardation will be obvious from birth or early infancy while mental illness may become manifested any time throughout the individual's life. Mental illness may be treatable; mental retardation is not.

Many forms of mental illness are attributed to defective genes. One of the more common forms, manic-depressive, is thought to result from a single gene defect, and is transmitted in some families by means of the X chromosomes.

Schizophrenia, the most common form of all psychiatric diseases, is at the same time the most misunderstood and the least understood illness to undergo genetic investigation. Schizophrenia is the name for an illness affecting the mind and personality of an individual in a way which is seldom completely resolved.

The variation in symptoms of schizophrenia is one of the factors that has made the illness difficult to diagnose. Now over 95 percent of schizophrenic patients are so diagnosed

according to a "nuclear syndrome" based on auditory hallucinations, thought disorders, and a loss of will.(8)

The lack of agreement on and interpretation of data makes the manner of inheritance remain unresolved. Like mental retardation, schizophrenia may be a collection of disorders with different causes and courses, but most behavioral scientists agree that biochemical factors play an important role in the illness.

According to Dr. E. Fuller Torrey, author of "Surviving Schizophrenia: A Family Manual," schizophrenia is not caused by childhood trauma, domineering mothers and/or passive fathers, or guilts, acts or failures of the victim.

Dr. Torrey states schizophrenia is: a real disease with concrete and specific symptoms, a result of flawed brain biochemistry, sometimes curable, and often has a genetic component. It may be treatable with specific psychotropic drugs, but is not responsive to treatment by psychotherapy.(9)

Schizophrenia strikes both sexes about equally, but the age of first admission to a hospital differs somewhat between the sexes, males showing symptoms first at a ratio of 4 to 1 in infancy, leveling out in the middle thirties, and women showing a 2 to 1 ratio of new cases by the late fifties.(4)

The frequency of schizophrenia in the general population is extremely common with an incidence rate in the 1 percent range. However, the frequency rate of schizophrenia among relatives of a person diagnosed with the disorder is decidedly higher than that for individuals in the general population;

the rates vary from between 2 and 46 times higher than the average risk for schizophrenia in the population. First degree relatives show a higher frequency of occurrence than second or third degree relatives. Children with both parents affected show a higher occurrence rate than those with only one affected parent.(10)

Evidence suggesting that genetic factors are involved in the causes of schizophrenia is shown by: family studies, twin studies, and adoption studies. In the family studies a comparison was made between the frequency of the disorder among relatives of affected persons and the base rate of the disorder in the general population. The significantly higher rate among relatives suggests a genetic factor to be a cause of the disorder, but the irregular pattern of inheritance does not support a particular method of transmission (i.e., by a recessive gene, a dominant gene, or a sex-linked chromosome).

Studies of schizophrenia among twins has been done to determine if schizophrenia is genetically induced. If the genetic theory is true a higher incidence of the disorder should occur between identical twins since their inherited genetic material would be the same. The incidence of the disorder between fraternal twins should be no higher than that of other siblings since the inherited genetic material is not identical. Studies show there is indeed a higher rate of occurrence between identical twins than among fraternal twins.

The most compelling evidence that genetic factors are involved in the causes of schizophrenia come from studies on adopted children. Independent studies by Heston (1966) in Oregon and Rosenthal (1968, 1971) in Denmark agree that the offspring of a schizophrenic parent have substantially the same risk for the disorder whether or not they are raised by the affected parent. Studies by Wender (1974) suggests that the experience of being reared in a schizophrenic home does not increase the risk for schizophrenia in an individual unless a genetic predisposition for the disorder is already present, and children of schizophrenic parents raised by their own parents or by adoptive parents show the same incidence of the illness.(10)

Despite the difficulties of methodology, the studies--family, twin, and adoption--all point in a common direction: persons with a family background of schizophrenia are more likely to become affected than those without family background of the disorder.

While the evidence does indicate schizophrenia is inherited two factors cast doubt: the mode of inheritance of the gene or genes involved in the cause of schizophrenia does not follow the simple Mendelian pattern, as do many traits and diseases in many different species, and the high frequency occurrence of schizophrenia in contrast with the low rate of other severe genetic diseases found in the general population.(4)

According to the 1979 Annual Report of Mental Health

Care in Oklahoma, alcohol and drug abuse was the leading cause of admission to state mental hospitals.(11) That fact makes it worthwhile to briefly mention the studies done on the genetic basis of alcoholism.

Studies involving the same methods as those for schizophrenia--family, twins, and adoptions--have arrived at much the same conclusion; evidence suggests alcoholism has some genetic influence.(12)

CHAPTER V

THE FUTURE OF GENETICS AND MENTAL HEALTH

Scientific studies, technological improvements, and medical research working together can now help answer questions expectant parents may have concerning the mental health of their unborn child. More than 3,000 gene determined diseases are known to exist, many of them associated with mental retardation. Most of these diseases can be diagnosed prenatally.(13)

The detection of genetic abnormalities in the fetal state may be done by amniocentesis. Amniotic fluid is withdrawn abdominally and the cell samples cultured and assayed for chromosomal abnormalities and biochemical defects.

Ultrasonography is a major tool for prenatal diagnosis. Among fetal abnormalities that can be visualized are anencephaly, hydrocephaly, fetal ascites, and renal agenesis.

Direct visualization of the fetus is possible by fetoscopy. The entire fetus may be scanned, looking for physical abnormalities that might be clues to the presence of a genetic syndrome.

Both amniocentesis and fetoscopy carry some risk of infection and a slight increase in the probability of spontaneous abortion.(14)

The human genome is estimated to contain 50,000 to 100,000 genes. In 1983, the chromosomal location of more than 350 of these genes was known.[6] Future medical advancements will see the mapping, cloning, and study of the human genes to understand more about the body's normal function at the molecular level. As more knowledge is gained at the molecular level gene therapy or transplant may become standard treatment for genetic disease.

The first successful gene therapy in a mammal was reported in 1984 to correct a genetic deficiency of a growth hormone in a mouse. Partial correction of an enzyme deficiency causing the disease Lesch-Nyhan by introducing a normal gene into human bone marrow cells growing in a culture has been successful.[14]

It is now possible to isolate radioactive probes for specific genes and to study these genes in cultured cells by using new techniques of molecular biology. Cell cloning has been a valuable help in the study of human disorders. More recently the use of specific enzymes to reproduce chromosome material has provided a major break through in the study of human inborn errors.

Genetic counseling, as to the probability of having affected children based on family background or the rate of incidence in the general population, may soon be outdated. It has been estimated each one of us carries six lethal genes in his genetic material. Carriers for many lethal genes can be detected. Carriers have lower than normal

levels of the enzymes which are produced by the specific mutant gene they carry. Is it possible somewhere in the future, perspective parents will be required to compare computer print-outs of their genetic make-up--which was determined before their birth--for harmful dominant and recessive genes before being allowed to parent children?

CHAPTER VI

CONCLUSION

Many forms of mental retardation have a genetic basis. Down Syndrome is the most common genetically induced retardation and is most often caused by an excessive amount of chromatin material.

Some forms of mental retardation are caused by gene mutation. Phenylketonuria (PKU) is the most common disease caused by mutant gene action which can result in mental retardation.

Studies on the causes of schizophrenia, the most frequently occurring form of mental illness, are inconclusive, but evidence indicates schizophrenia has a genetic basis. According to the studies presented, environment plays a small role in the etiology of schizophrenia.

Although there is a persistent impression that labeling a disease as genetic is labeling it as incurable, actually many genetic disorders can be treated with a reasonable degree of success--sometimes by simply altering the diet. In some genetic disorders the victims may be at risk only under certain environmental conditions.

Parents can be less anxious about the mental health of their unborn child. Tests can now reveal mental health

as well as physical health with a reasonable degree of accuracy.

The future of genetics to insure good mental health is both frightening and exciting. A lot of moral and legal questions have to be answered before the knowledge that is now available to us can benefit victims of genetic based poor mental health.

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Scope of Study: The major concern of an expectant mother is not the sex, but the health--particularly mental health--of the unborn child. The purpose of the study was to provide a layman's understanding of the basic principles of heredity and the genetic causes of mental retardation and mental illness. An attempt was made to determine if heredity does influence mental health. The incidence rate for specific types of mental retardation is included as well as an explanation of the difference between inherited mental retardation and mental illness. There is a brief description of prenatal tests for chromosomal abnormalities which may lead to mental retardation, the role of the environment on mental illness, and a look into the future of genetic studies on mental health.

Findings and Conclusion: Since 1934 when the relationship between Phenylketonuria (PKU) and mental retardation was discovered there have been other mental diseases linked to inherited faulty biochemical processes. Currently there are about 1350 single-gene determined diseases, many of them associated with mental retardation. Although many forms of mental illness are attributed to defective genes, the study of schizophrenia--the most common form of all psychiatric diseases--is still inconclusive and it remains the least understood illness to undergo genetic investigation.

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