

SIB

SIBLING INFORMATION BULLETIN

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AHRC New York City

Fall 2000

GENETICS AND GENETIC COUNSELING

INTRODUCTION:

This bulletin will provide you with a brief overview of genetics and genetic counseling, several composite sibling-group summaries of our experiences with genetic counseling, and a list of genetic counseling resources available to you and other family members. We hope it will be a good introduction to a subject important to many of us.

I. GENETICS OVERVIEW

Genetics is the scientific study of how our physical, biochemical, and behavioral traits are transmitted from one generation to the next; scientists engaged in such study are known as geneticists. Genes, the basic units of hereditary material, are composed of proteins and nucleic acids. Each of our cells contains some 100,000 genes which are located on intertwined ladder-like structures called chromosomes. Each parent contributes half, or twenty-three, chromosomes to their offspring.

“Genetic” abnormalities fall into three categories: those arising from

disorders in the genes - more than seven thousand of these disorders have been identified so far, although most do not involve developmental disabilities; chromosomal disorders, in which there are too many or too few chromosomes; and multifactorial disorders, which have both genetic and environmental causes.

While our siblings’ disabilities may be due to genetic abnormalities, most are probably not genetic in origin. For example, maternal medicine and drug use, prematurity, or neonatal illness may all lead to developmental delays. Genetic and non-genetic factors together account for the estimated 3% of all babies born with developmental disabilities.

Scientists have determined that genetic factors account for up to sixty percent of severe mental retardation; environmental factors such as maternal medicine and drug use, exposure to chemicals and nutritional imbalances are more likely to be causes of mild mental retardation.

The “Human Genome Project,” which has now delineated or mapped the location of all of the genes on our chromosomes holds great promise for future work in identifying the gene or genes of disorders whose causes are unknown. Ultimately, though not in the near future, “gene therapy,” or substituting “healthy” genes for abnormal ones may be possible. ■

II. GENETICS COUNSELING

Genetic counselors are health professionals with specialized graduate degrees and experience in the areas of medical genetics and counseling. As members of the health care team, they:

- ◆ Provide information about your risks for giving birth to a baby with a genetically-based disorder. You may wish to embark on genetic counseling as a single person or with your spouse, to try to ascertain the risk factors each of you brings to your potential offspring
- ◆ Prepare you for the kinds of information you may receive from your test results
- ◆ Give you support regarding your options once the test results are known

To understand your risks, the genetic counselor may want to obtain your family history, your genetic profile obtained from blood testing, and other, life-style data. The most important issue the counselor addresses is whether your sibling's disability is a genetic one, so it is helpful for your sibling to be involved in the process. If the results indicate a genetic cause for his/her disability, your blood will be tested for the same genetic abnormality, as well as for other conditions for which you may be at risk.

Genetic counseling should also be sought if you are pregnant and considering amniocentesis or CVS (chorionic villus sampling). Counselors will help you understand the benefits and limitations of these procedures. ■

III. COMPOSITE SCENARIOS FROM SIBLING GROUPS

Amy and Joe's brother John, the oldest, was born with mental retardation. The family always believed John was born with mental retardation because his mother had had several serious illnesses with high fevers during her pregnancy with him. Amy is a direct care worker at a local agency for people with developmental disabilities, and studying for a bachelor's degree. A recent topic in class was genetic and non-genetic causes of mental retardation, including something called fragile X syndrome, the most common heritable cause of mental retardation. Hearing about the characteristic appearance of men with this disorder, Amy suddenly became aware that John resembled the profile her professor presented. It had never occurred to her that John might have something he *inherited*. If John did have fragile X, then she and Joe might be carriers, and subsequently transmit fragile X to their respective offspring.

Amy couldn't stop thinking about the possibility that John had fragile X syndrome. She had looked forward to getting married, and she loved kids. What did this possibility mean for Joe and her? Now what?

Amy ultimately decided she needed to know more. After looking at websites and resource guides (see resources on page 4), she decided her intuition might be based in fact. She talked her mother into contributing blood, and her brother John, too—they didn't have any other relatives still living. To Amy's shock,

the genetic counselor concluded that John didn't have fragile X syndrome, but he *might* have a so-called multifactorial kind of mental retardation resulting from the effects of several genes together with non-heritable factors. Amy and Joe might never discover exactly why John was mentally retarded. It might be just as the family believed or his disability might be due to other causes. It might never be clear whether Amy or Joe were "carriers"—or, exactly what, if anything, they could pass along to their children. Her (and Joe's) future decisions would have to be made with some uncertainty. And yet, Amy could not forget that sinking feeling she had when she thought she could possibly have a child like John.

Jamie was 41 when she discovered she was pregnant. She was thrilled because she'd always wanted a child. This was her parents' first grandchild and they were thrilled too. Jamie took good care of herself—no drinking, smoking or caffeine and she followed all of her doctor's advice on supplements and appropriate levels of activity. In the third month of her pregnancy, Jamie began to catch her mother watching her out of the corner of her eye with a worried look. Jamie soon became upset with her mother's growing anxiety and confronted her. Jamie's mother admitted she was worried because her brother Leon had had mental retardation. Suppose Jamie was carrying a child with a developmental disability?

As a child, Jamie had known and

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loved her uncle Leon, who, as far as anyone knew, had mental retardation of an undetermined origin. But it had not occurred to Jamie that the fetus she carried would be anything but healthy, in part because she was healthy and had undergone whatever tests her doctor had recommended, and in part because she and her husband Justin, wanted a baby so very badly. Her mother told her that not even amniocentesis could guarantee a baby without any disability--and gave her quite a bit of information on what it was like to have a person with mental retardation in the household. Jamie and Justin listened, trying not to catch her anxiety and to respect her perspective. They again talked to their doctor who explained the odds of their baby's having a disability, including one related to Leon's condition. The doctor gave them a referral to a genetic counseling center nearby, even though Jamie was already pregnant. After a tremendous amount of discussion and further thought, Jamie and Justin decided not to have genetic counseling. They knew the tests they had elected weren't perfect; they also decided that although genetic testing might shed additional information on their risks, it wouldn't affect their decision to have the baby. Jamie's mother couldn't understand their choice to have a baby without doing everything possible to know in advance whether there was "something wrong," and said she would not help Jamie and Justin if their baby should have a developmental disability.

Anne's sister Andrea has fragile X syndrome. Their family was one of those originally tested for the genetic disorder, and Anne can tell you when she found out she was a

carrier. She remembers because she felt her whole world collapsing around her, not the least because she was already married to Norman, who, while tolerant of Andrea, was convinced that Andrea's mental retardation had nothing to do with Anne and him. Anne so much wanted to avoid all the emotional issues related to being part of a family with a genetic disorder! And her love for, and knowledge of "the way Norman is" made the whole situation more challenging. Genetic mapping of her family through blood work and their histories did not provide comfort--except, as her father said, at least she knew "in advance."

Anne still remembers the night she told Norman she was a carrier. Norman, who wanted a son, wasn't too keen on talking about any of this, but equally, wasn't inclined to have a baby he knew ahead of time would be disabled. Anne did what she could--lined up people Norman could talk with and thought through her choices and what she wanted. Her mother was willing to talk to Norman because she, herself, hadn't known--how different the world was when you don't know!

In the end, however, when Anne got pregnant, she went through the tests alone, and when it showed the fetus was affected, decided to terminate the pregnancy. It wasn't that her family wasn't involved, and it wasn't that there weren't disagreements about what Anne should do. But Anne had felt pretty much alone with the decision, which after all, had to do with *her* side of the family. She reassured herself with the knowledge that she could get pregnant in the future and have a completely normal baby. ■

IV. SIBLINGS SPEAK FROM EXPERIENCE

From Ms. X: My experience with genetic counseling was a necessary one despite the fact that it did not provide me with a definite and concrete answer.

As a young woman with two mentally retarded brothers, I wanted to obtain information about the risk of my having a child with mental retardation. So, I went for genetic counseling at a New York hospital where my family history and a blood test were taken. It was determined through the blood test that my brothers did not have fragile X syndrome. This condition, which is X-linked (ed. note: carried by women) and affects males more than females, is the most common heritable form of mental retardation.

The genetic counselor and doctor at the hospital told me that since fragile X syndrome was ruled out, they did not know what had caused my brothers to have mental retardation. They informed me that the cause could be recessive genes, X-linked, or just "an act of God" with the reason unknown. Since the cause was not determined, they were not able to give me a specific prediction of what my chances of having a child born with mental retardation would be. I was told to be in contact with them periodically, since new research on the causes of mental retardation are constantly being done.

Despite the fact that I am still not certain about what the actual risk is

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of my having a child with mental retardation, I know that I have done all that I can to obtain a better understanding of the causes of my brothers' mental retardation. For me, even being told that the cause is "not known" is still an answer; otherwise, I would automatically assume a genetic cause.

I recommend genetic counseling to siblings of individuals with disabilities. Genetic counseling can help increase your knowledge of present and potential risks. It can provide you with an opportunity to make an informed decision about pregnancy. ■

The following is from "**Good News--Genetic Counseling**," a 1985 article in the SIBLING INFORMATION NETWORK: Newsletter, published by the Department of Educational Psychology, The University of Connecticut, Storrs, by Bonnie Cohen, formerly both an AHRC board member and chair of the Sibling Committee. Both Bonnie and her handicapped brother gave blood samples from which "chromosomal prints" were produced, and no genetic abnormalities detected. It was felt that her brother's retardation was due to "brain damage," and that her chance of bearing a retarded child was the same as anyone's. Bonnie's feelings at the conclusion of this genetic counseling experience:

"By isolating my brother's handicap in this way, I feel much stronger and more normal. I believe in my separateness more than ever, and no longer dwell in the shadow of the chronic grief which encompasses so many families with developmental disabilities." ■

GENETICS/GENETICS COUNSELING WEBSITES:

The **ARC of the United States:**

www.thearc.org

A national organization "of and for people with mental retardation and related disabilities and their families;" the site has lots of interesting articles on genetics.

The **National Society of Genetic Counselors:**

www.nsgc.org

This site has an international directory of genetic counselors and links to disability-related sites.

Genetics Education Center, University of Kansas Medical Center:

www.kumc.edu/gc/support

Information site on genetic and rare conditions, includes information about support organizations. ■

RESOURCE GUIDE FOR GENETIC RESEARCH AND COUNSELING

BRONX

Lincoln Medical and Mental Health Center, Division of Genetics
718-579-5295

Montefiore Medical Center, Division of Reproductive Genetics
718-405-8150

Montefiore Medical Center, Center for Congenital Disorders
718-920-4300

BROOKLYN

**The Brooklyn Hospital Center, Departments of
Obstetrics/Gynecology and Pediatrics Reproductive
Genetics/Pediatric Genetics** 718-935-7410

Interfaith Medical Center, Brooklyn Jewish Site 718-935-7410

Maimonides Medical Center 718-780-3246

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Sibling Information Bulletin

IS WRITTEN BY THE SIBLING
COMMITTEE OF AHRC

NEW YORK CITY
200 Park Avenue South
New York, NY 10003
212-780-2500
www.ahrcnyc.org

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SUNY Health Science Center at Brooklyn, Department
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Harlem Hospital 212-939-1701

Metropolitan Hospital Center, Department of
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The Mount Sinai Medical Center, Department of
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Presbyterian Hospital in the City of New York,
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St. Luke's-Roosevelt Hospital, Division of Genetics
212-523-5895

St. Vincent's Hospital and Medical Center of New
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MAILING LIST: Do you have a family member, or know someone with a developmentally disabled sibling who would benefit from being on the mailing list? Send their name and address to AHRC - 200 Park Avenue South, NY, NY 10003.

Name _____

Address _____

City _____ State _____ Zip _____

Phone _____

AHRC's Sibling Program sponsors a monthly Adult Sunday Sibling Support Group. For the schedule and information, call Dr. John Fiorello at (212) 780-2592.

Only \$10 Will Make You a Member of AHRC

QUEENS

City Hospital Center at Elmhurst, Department of Pediatrics 718-334-5510/5111

Long Island Jewish Medical Center, Schneider Children's Hospital 718-470-3010

Queens Hospital Center 718-883-4676

STATEN ISLAND

Institute for Basic Research in Developmental Disabilities
718-494-5211

LONG ISLAND

The Genetics Center 516-862-3620

Nassau County Medical Center 516-572-3148/3391

North Shore University Hospital, Division of Child Development and Human Genetics 516-365-3996

SUNY at Stony Brook, Genetics Division, Department of Pediatrics 516-444-2790

Winthrop University Hospital 516-663-2657

WESTCHESTER

Regional Medical Genetics Center, Inc. 914-347-3010

UPSTATE

Children's Hospital of Buffalo Division of Genetics
716-878-7530

University of Rochester School of Medicine
716-275-3304

SUNY Health Science Center at Syracuse, Regional Genetics Program 315-464-7610

Binghamton Genetic Counseling Program
607-724-4308

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New York, NY 10003

212-780-2500

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