PERSPECTIVES IN GENETIC COUNSELING

NATIONAL SOCIETY OF GENETIC COUNSELORS, INC.

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RESOURCES FOR PARENTS & FAMILIES

Trisomy 18: A Book for Families. Beth Fine, Karen Greendale, Kris Holladay, and John Carey, 1982.

Trisomy 18: A Book for Families is an attractive, well-organized, 41-page pamphlet--complete with photographs, diagrams, references, resources, and a glossary--about this rare and devastating condition. It is coauthored by two genetic counselors, a physician, and the coordinator of a support group for trisomy 18 and 13. Although this resource has strong points and may be helpful for some families, its major drawback is that it presents an unrealistic picture of the prognosis for trisomy 18.

The book begins with a clear and concise description of the features characterizing trisomy 18 and a good definition of the term "syndrome." One does not, however, get the impression that trisomy 18 is a very severe, most often fatal, condition where the few survivors exhibit extreme developmental delay and mental retardation.

The clinical description of trisomy 18 is followed by a section on chromosomes, which is too detailed and quite complicated. The authors spend a great deal of time differentiating among trisomy 18, trisomy 18 mosaicism, and partial trisomy 18 and explaining how each arises. I question both the content of this section and its placement on pages 4-13 of the brochure. A lay parent who finds this information confusing may simply read no further and discard the pamphlet. Parents of a newly-diagnosed infant with trisomy 18 experience many emotions and are mourning the loss of the normal child they had anticipated throughout the pregnancy. They are not likely ready for this type of information. A shorter, simpler explanation of the chromosomal basis of trisomy would certainly suffice. The recommendation that the family meet with a genetic counselor to discuss the family's situation is good, as is the statement that several sessions may be required to comprehend this information fully.

One-third of the way through the booklet, in the section on the meaning of the diagnosis, one gets the first hint that most babies with trisomy 18 die before one year of age; but the statistic that is quoted is that 50% of these infants die before the age of six months. The authors then proceed to deal with the anger, disbelief, and guilt that parents universally feel. Before the information about early death can be thoroughly digested, the reader is swept into a discussion about bringing the baby home, learning special feeding and monitoring techniques, and using only a regular pediatrician, rather than a specialist, for medical care. The discussion assumes that major defects such as congenital heart disease or cleft lip/cleft palate will automatically be treated successfully. The pamphlet ignores the fact that most infants with trisomy 18 will never be well enough to leave the special-care nursery or the hospital.

There is a gentle suggestion that developmental delay is part of the picture for trisomy 18. The authors do not mention that there are very few long-term survivors or that the retardation can be severe. Hearing tests and infant stimulation programs are recommended, and one can quickly forget the ominous prognosis that is the general rule for trisomy 18. The exception to the rule, rather than the rule itself, is stressed.

The discussion of family dynamics is certainly a strength of this publication. That discussion emphasizes that there are no right or wrong feelings or courses of action, and that each family should do what is comfortable for it at any particular time. The authors deal with real problems that trisomy 18 families confront--such as bonding with the baby, whom to tell, how much to tell, and informing the other children--as well as with problems that only a few families face--such as respite for the family caring for a surviving child at home, foster care, and institutionalization. The issue of life support and heroic care is especially important, and the parents' role in this difficult decision is addressed well. The authors state that the baby may never leave the hospital alive, and that this is very hard to acknowledge and accept.

Quotes from parents are used throughout the pamphlet; they give a new parent of a child with trisomy 18 the feeling that he or she is not alone, and that this has happened to other people from all walks of life. The photographs also serve to reaffirm that this happens to real people and real families. Most of the quotes tend to be encouraging and optimistic and are from families whose affected child has given them new insights into life. Unfortunately, that may not be true for every family, and a family that is having difficulty accepting and coping with the diagnosis and the affected child may be made to feel unusual or abnormal.

The roles of the genetic counselor and other medical professionals are discussed throughout the booklet. Families are encouraged to ask any and all questions and to meet with various specialists as needed. The discussion of amniocentesis, which appears at the end of the publication, is appropriate and well done. The list and description of support groups (with contact persons and telephone numbers) and references (with addresses and prices) should be useful for families, who can refer to them at an appropriate time.

Although families that have a child with trisomy 18 often request written information about the disorder, I have reservations about recommending this book to them. That is due to its unrealistic emphasis on the very few babies who survive and its confusing discussion about chromosomes. Any genetic counselor contemplating the use of this publication should read it several times and should feel comfortable with it before distributing it to patients.

Trisomy 18: A Book for Families is available for $2 per copy from the March of Dimes Birth Defects Foundation, Southeast Nebraska Chapter, Boardwalk 11917 Pierce Plaza, Omaha, NE 68144-598.

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The Cri-du-Chat Syndrome. Louise E. Wilkins, Judith A. Brown, Barry Wolf, and Walter E. Nance, 1982.

This concise booklet on cri-du-chat syndrome was prepared by professionals at the Medical College of Virginia who have worked with 86 children with the disorder. The booklet combines information obtained from the study of those children with descriptive information on chromosomes, chromosome abnormalities, and the 5p- syndrome.

The booklet begins with a brief description of cri-du-chat syndrome. The next several pages are devoted to a description of chromosomes, an explanation of the specified chromosome abnormalities that lead to the syndrome, and an explanation of terminal deletions, interstitial deletions, inherited unbalanced translocations, and *de novo* unbalanced translocations. The features of the syndrome are discussed in general terms. There is also a section, supplemented with photographs, devoted to the changing physical features of these children as they age.

The authors review birth defects that may be associated with cri-du-chat syndrome and describe the findings from their study of the 86 children in this population. Several pages are devoted to the range of intellectual development in these children, and to suggestions on how early special education influences the developmental level of the affected child. An excellent section on home care and residential placement emphasizes the importance of the individual family decision involved here.

Genetic recurrence risks are discussed throughout the booklet. A very brief and somewhat incomplete discussion of prenatal diagnosis closes the narrative portion of the book. Finally, there is charting space where parents can record their child's progress. There is a form that has room for chromosome findings on the affected individual and parents, along with information on hospitalizations, measurements, illnesses, and developmental milestones. There is also a section to note results from psychological testing and schooling.

Overall, this booklet is quite complete, particularly in its discussion of chromosomes, karyotype preparation, and the different ways that the cri-du-chat syndrome can arise chromosomally. There is also a very good mix of general information and results from the study noted above. Most of the children were raised at home, so that the developmental levels appear to be higher than those usually presented in the literature. Almost all of the children in the study group were under age 10; information on older children who may not have had the benefit of infant stimulation and home rearing is therefore not included in this sample.

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This is a useful resource for families; it reiterates information parents may have learned in a genetic counseling session. It offers families hope, yet is realistic about the delayed development of children with the cri-du-chat syndrome. The amniocentesis section should have been expanded to mention the alternatives if any abnormality is found. Although the discussion of chromosome analysis is a bit detailed for most parents, that certainly does not detract from this small, but useful booklet.

The Cri-du-Chat Syndrome is available from The Department of Human Genetics, 11th and Marshall, Box 33, MCV Station, Medical College of Virginia, Richmond, VA 23298.

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Neurofibromatosis: A Primer for Patients and Families. Vincent M. Riccardi and Susan H. Valenta, 1982.

This 16-page booklet is written in question-and-answer format. Dr. Riccardi and Ms. Valenta have done a good job discussing an extremely variable disorder in a manner that is not overly threatening to the patient and his or her family. Neurofibromatosis (NF) and its many manifestations are described in detail. Graphic photographs of cafe-au-lait spots, neurofibromas, as well as CAT scans showing optic gliomas, neurofibromas, and malignant schwannomas, add to the descriptive nature of this book. The booklet emphasizes the variability of the disease, explaining that there is no consistency within or between families, and there is no way to predict the course of the disease in an individual. The authors describe clinical features such as cafe-au-lait spots, neurofibromas, Lisch nodules, and various tumors, along with non-specific findings such as speech impediments, seizures, scoliosis, and school performance problems. Treatments for some of those problems are mentioned briefly in the booklet.

Subsequent sections include discussions of the genetics of NF, and of the social and emotional impact of NF on families. The authors emphasize throughout that many individuals affected by NF lead normal lives. They stress as well the need for a good relationship with a physician so that the family can be offered the best possible treatment and the best possible examinations for prevention. In addition, genetic counseling is emphasized as crucial in prevention and in family planning. The booklet concludes with a discussion of the Baylor Neurofibromatosis Program.

I found this booklet useful with families, because it is a bit more detailed than the flyers available from the National Neurofibromatosis Foundation or from the National Institutes of Health. If a family is particularly interested in participating in the Baylor program, the genetic counselor could help arrange an appointment. The booklet is nicely designed and is easy to read.

The booklet is available by writing to Dr. Vincent M. Riccardi, NF Program, Baylor College of Medicine, 1200 Moursund, Houston, TX 77030. A single copy of the booklet is available by making a donation of $3.00 to $5.00 to the Baylor NF Program. Inquiries about multiple copies are encouraged.

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POSITIONS AVAILABLE

Genetic Associate: A position is now available for a genetic associate in the Human Genetics Program, Tulane University Medical School, as counselor and coordinator of active general genetics clinics. Master's in genetic counseling, board certified or ligible is preferred, but all applications will be considered. For ıformation call or write: Emmanuel Shapira, MD, PhD, Human Genetics Program, Hayward Genetics Center, Tulane University School of Medicine, 1430 Tulane Avenue, New Orleans, LA 70112, phone: (504) 588-5229.

Genetic Associate: The Department of Clinical Genetics of the Children's Medical Center, Tulsa, Oklahoma, has a position available for a genetic associate. Primary responsibilities will include coordination of satellite clinics in eastern Oklahoma, genetic counseling, and genetic education of the medical and lay communities. Applicants must have a master's degree in genetic counseling and be board certified or eligible as a genetic counselor with the American Board of Medical Genetics. Submit curriculum vitae and references to: Nancy Carpenter, PhD, Children's Medical Center, 5300 East Skelly Drive, Tulsa, OK 74135, phone: (918) 664-6600.

Genetic Counselor: One position in genetic counseling is available at the University of Illinois-Peoria. For further information applicants should contact: Reuben Matalon, MD, PhD, University of Illinois, Department of Pediatrics, 840 South Wood Street, Chicago, IL 60640, phone: (312) 996-6714.

Genetic Counselor: A new position is available immediately at the University of Kansas Medical Center, Kansas City, Kansas. Depending on interests and abilities, responsibilities will include general genetics serving pediatrics and internal medicine, clinical research, education programs and workshops, specialty clinics (cystic fibrosis, cranio-facial, etc.), five statewide outreach clinics, and family support groups (Huntington's disease, Little People, etc.). Prefer American Board of Medical Genetics certified or eligible. Contact: Deborah Collins, MS and Neil Schimke, MD, Division of Metabolism, Endocrinology, and Genetics, University of Kansas Medical Center, Rainbow Blvd., at 39th Street, Kansas City, KS 66103, phone: (913) 588-6043.

Genetic Counselor: The Genetics Center of Southwest Biomedical Research Institute in Tempe (Phoenix) has an immediate opening for a full-time genetic counselor. Primary responsibilities include genetic counseling, participation in coordination and administration of clinic, specialty clinics, genetics education for medical and lay communities, and participation in research projects. Applicants must have an appropriate master's degree, for example in human genetics or genetic counseling, and be board eligible or certified as a genetic counselor with the American Board of Medical Genetics. Please submit current curriculum vitae to: Judith Allanson, MB, ChB, MRCP, Associate Director of Clinical Genetics, The Genetics Center of Southwest Biomedical Research Institute, 123 East University Drive, Tempe, AZ 85281.

Genetic Counselor: A position is available immediately with the State Division of Health Services, Greenville, North Carolina. This is a community-based position. Responsibilities include community education, coordination of five satellite genetics clinics, assisting health providers with case identification and referrals, providing initial and follow-up genetic counseling, and acting as a liaison between the medical genetics center in Greenville and local communities within that region of the state. A master's degree in genetic counseling is required. Salary commensurate with experience. Contact: Elizabeth G. Moore, Division of Health Services, P.O. Box 2091, Raleigh, NC 27602, phone: (919) 733-7437.

Genetic Counselor: The Genetics Unit, University of Colorado Health Sciences Center, Denver, has a position available 1 May 1984 for a genetic counselor/regional genetic counseling program coordinator. Responsibilities include coordination of rural clinics, periodic travel to rural clinics with MD-geneticists, genetic counseling, participation in post-graduate education of health professionals, and public education. Candidate must be board eligible/certified by the American Board of Medical Genetics and have at least 2 years experience in a general genetics clinic. Salary is based on experience. Send resume to: Eva Sujansky, MD, director, Genetics Unit, Box B-160, University of Colorado Health Sciences Center, Denver, CO 80262, phone: (303) 493-8808.

Genetic Associate: The Department of Medical Genetics/Crippled Children's Division of the Oregon Health Sciences University will have a vacancy in the summer of 1984 for a genetic associate at the Regional Services Center in Eugene. The primary emphasis of the position involves clinical and educational activities, including clinic coordination, general genetic and prenatal counseling, and both public and professional education. Applicants must have a master's degree and be board eligible/certified by the American Board of Medical Genetics. Previous clinical experience desired. Send applications and curriculum vitae to: Jonathan Zonana, MD, Crippled Children's Division, Clinical Services Building, University of Oregon, Eugene, Oregon 97403, phone: (503) 686-3575.

JOBS HOT-LINE NUMBER
Linda Nicholson: (302) 651-4234

NSGC NEWS

NSGC Members Prefer To Hold Annual Education Conference In Conjunction With The American Society Of Human Genetics Meetings

The first four NSGC National Education Conferences (1981-1984) have been held in conjunction with the March of Dimes Birth Defects Conference, and the society received financial support from the March of Dimes for those meetings. March of Dimes' policy is to fund a project such as ours for a maximum of five years. We have been informed that we may receive a small grant for the 1985 meeting, but that we should consider other sources of funding for future meetings. The NSGC conferences have grown from about 125 participants in 1981 to 200 in 1983.

At the 1983 NSGC business meeting in Norfolk, Virginia, several members expressed concern about limited travel funds for conferences. Many felt that if travel budgets allowed one meeting per year, the American Society of Human Genetics (ASHG) meeting was the meeting of choice. However, that creates a dilema for NSGC members who wish to attend the NSGC meeting as well. Accordingly, the NSGC education committee conducted a survey to determine the membership's preference on when to hold the meeting. Approximately 60 percent of the membership responded. Seventy-nine percent preferred to hold the meeting in conjunction with the ASHG meetings, 11 percent elected to hold it prior to the Birth Defects Conference, 4 percent wished to hold the meeting independently, and 6 percent had other suggestions. The reason for changing the meeting was, again, financial. Several respondents preferred that the meetings still be held in conjunction with the Birth Defects Conference, because of the busy schedule of the ASHG meetings.

In response to the request of the general membership, the 1985 NSGC Education Conference will be held on 7,8 October in Salt Lake City, Utah. The theme of the conference will be: "Strategies in Genetic Counseling: Religious, Cultural, and Ethic Influences on the Counseling Process." Barbara Bowles Bieseckr has agreed to chair the conference. Anyone interested in working on the conference should contact her at: (608) 262-1007.

At this time, the education committee is exploring alternative sources of funding to help support the 1985 meeting. The committee will be working with the local arrangements committee for ASHG to maximize resources available for the conference. Anyone who has suggestions for possible funding sources or ideas for workshops, speakers, or other program items should contact Beth Fine at: Clinical Genetics Center, Childrens Memorial Hospital, 8301 Dodge Street, Omaha, NE 68114, (402) 390-5488.

An Opening on the Staff of *Perspectives*

The editorial staff of *Perspectives in Genetic Counseling* seeks an editor for a new column on case reports in genetic counseling. Please see *Perspectives,* Vol. 5, No. 4, December 1983, for a description of the case reports. Interested persons should write: J. D. McInerney, *Perspectives,* BSCS, The Colorado College, Colorado Springs, CO 80903, or phone: (303) 473-2233, ext. 736.

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