THE STUDENT WITH A GENETIC DISORDER

A project of the
Pacific Northwest Regional Genetics Group
Child Development and Rehabilitation Center
Oregon Health Sciences University, Portland Oregon

THE STUDENT WITH A GENETIC DISORDER

Educational Implications for Special Education Teachers and for Physical Therapists, Occupational Therapists, and Speech Pathologists

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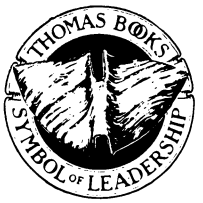

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INTRODUCTION

This book has a two-fold purpose. The first is to acquaint special education teachers, therapists, child development specialists, and other health professionals with general genetic concepts and the physical characteristics, learning profiles, and health and related problems of children with certain specific genetic conditions. The second is to suggest educational and therapy interventions for these children in classrooms and child development programs.

In this text syndromes are grouped with others that share common features. These conditions were chosen because they have a significant impact on the child's education. There is a paucity of material available on many of these rare genetic conditions. At the same time individuals and their families are increasingly demanding knowledge about their particular genetic condition. Educators find an increasing need to communicate intelligently with parents, health professionals, and other school professionals about the multiple ramifications of a specific diagnosis.

Although individuals with a specific genetic diagnosis share common characteristics, each child is unique. It is the child in the classroom who is to be considered, not the diagnosis. Each student needs the same sensitive caring approach with recognition of individual strengths and weaknesses. An important role of the educator and school health professional is to enhance the development of the child through knowledge of the physical, intellectual, and psychosocial impact of their diagnosis. By having adequate information, educational programs can be modified to fit the needs of the child, affecting the child's ultimate adjustment and functioning.

The text begins with a review of basic genetic principles and then discusses the characteristics of a genetic syndrome. It gives general information that is appropriate to many conditions: the elements of a barrier-free school, the role of the therapists, and when to consider a referral to a genetics clinic. Over forty genetic disorders are then described and discussed within general diagnostic categories. Of necessity, these descriptions are general and it is recognized that few students meet all of the diagnostic criteria for any one syndrome. Of even greater importance, the uniqueness and individual strengths and characteristics of each individual are clearly recognized. There

is a glossary defining many words frequently used and found in the medical literature, other suggested readings and references. A school checklist to help the child with special needs, a list of indications for referral to a genetics clinic, and a list of resources can be found in the Appendix.

This book is not designed to help determine a clinical diagnosis. It is to give an overview and to be used to answer general questions about the educational and therapy management of these students. On occasion, it may be a guide when parents have questions as to whether it is appropriate to refer their child to a genetics clinic for an evaluation. Technology is advancing at such a rapid rate that procedures that were in the test stages only months ago may be a plausible option at this time. Most states have medical centers where children with genetic disorders can be diagnosed and where families can receive help with specific questions.

The suggested educational and therapeutic interventions have been compiled from a variety of sources which include the medical literature, recommendations from specific parent and voluntary support groups, and from review of the educational, occupational, physical, and speech therapy records of the Child Development and Rehabilitation Center of the Oregon Health Sciences University, Portland, Oregon, and the Child Development and Mental Retardation Center of the University of Washington Medical Center, Seattle, Washington.

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The Pacific Northwest Regional Genetics Group (PacNoRGG) is one of ten regional networks under the umbrella of the Council of Regional Networks for Genetic Services. It is a consortium of providers and consumers of genetic services serving the states of Oregon, Washington, Alaska, and Idaho.

One of the primary goals of PacNoRGG is to promote professional education concerning genetic disorders. It is the partial fulfilling of this goal that has given the impetus for writing this resource book. The authors are from the staff and faculty of the Oregon Health Sciences University, University Affiliated Program, Portland, Oregon; the University of Washington School of Medicine and University Affiliated Programs, the Swedish Hospital Medical Center, and the Washington State Department of Health, Seattle, Washington; and from the Yakima Valley Community College, Yakima, Washington. This book could not have been completed without the encouragement and cooperation of the staff and faculty of these institutions.

Physicians, educators, and health professionals who have spent many years working intimately with children who have the conditions described in the text have carefully reviewed individual chapters. We cannot thank them enough for their thoughtful comments, added insights, and endless patience as they read and reread these pages. These helpful reviewers include: Thomas Bird, MD, Peter Byers, MD, Vanja Holm, MD, C. Ronald Scott, MD, J. Clifford Sells, MD, Robert Steiner, MD, Cristine Trahms, MS, University of Washington; Sarojini Budden, MD, Stephen LaFranchi, MD, Everett Lovrien, MD, Ellen Magenis, MD, Victor Menashe, MD, Rodney Pelson, PhD, Judi Tuerck, MN, Jonathan Zonana, MD, Oregon Health Sciences University; Jacob Reiss, MD, Kaiser Permanente, Northwest Region, Portland, Oregon; and Jay Rymeski, MEd, Current Communications, Portland, Oregon. Virginia Drawz, MA, from the Oregon Health Sciences University, and Jo Hoag, Marysville School District, Marysville, Washington gave many helpful comments on the Educational Implications addressed in each chapter. Susie Ball, MS, Central Washington Genetics Program, Yakima, Washington completed the final proofreading.

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THE STUDENT WITH A GENETIC DISORDER

Chapter I

BASIC GENETIC PRINCIPLES

*Genes** are the smallest unit of the hereditary material *deoxyribonucleic acid* or *DNA*. Genes are the blueprints for directing growth and development. Thousands of genes are packaged on chromosomes, like beads on a string. *Chromosomes* can be visualized under a microscope with special techniques, but genes cannot be seen with current technology. We receive half our chromosomes, thus half our genes, from each parent.

Chromosomes are found in all body cells except mature red blood cells. Within the nucleus of the cell are 46 chromosomes, or 23 pairs; 23 coming from the mother and 23 coming from the father. Of these, there are 22 distinct pairs called *autosomes,* and one pair called the *sex chromosomes.* In the laboratory the autosomes are arbitrarily identified and numbered according to their length, point of constriction and banding pattern. The sex chromosomes are also identified but given letters.

Females have two X's and males one X and one Y. The genetic difference between men and women lies in the sex chromosomes, the Y resulting in a male.

When new body cells are made, each chromosome goes through a process of making a copy of itself. The cell divides with each chromosome going into a new cell. This process, called *mitosis,* is similar in all living organisms and continues throughout life in all dividing cells.

This process differs in the production of the egg and sperm cells. The normal 46 chromosome cell reduces to a 23 count through a two-step reduction called *meiosis.* The egg has 22 autosomes and one X and the sperm has 22 autosomes and either an X or a Y chromosome. If the sperm with an X joins with the egg cell, a girl is conceived, if a sperm with a Y joins the egg, a boy is conceived. When the egg and sperm unite at conception the total complement of 46 chromosomes is reached and the fertilized egg starts immediately to divide by the process of mitosis and becomes the developing baby.

**Italicized words are found in the glossary.*

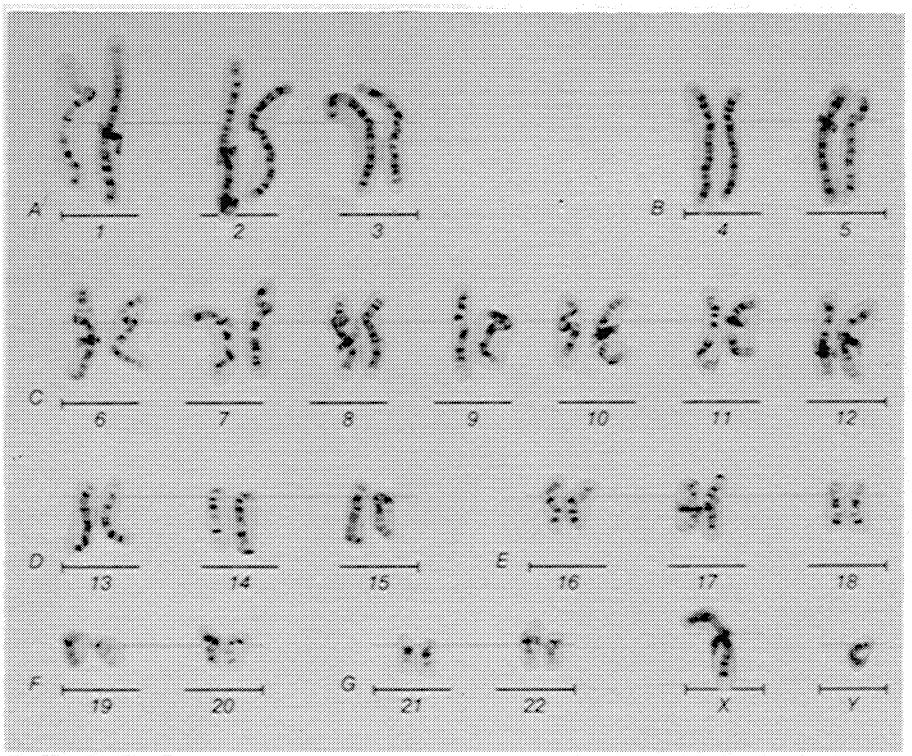

Figure 1. Normal male karotype.

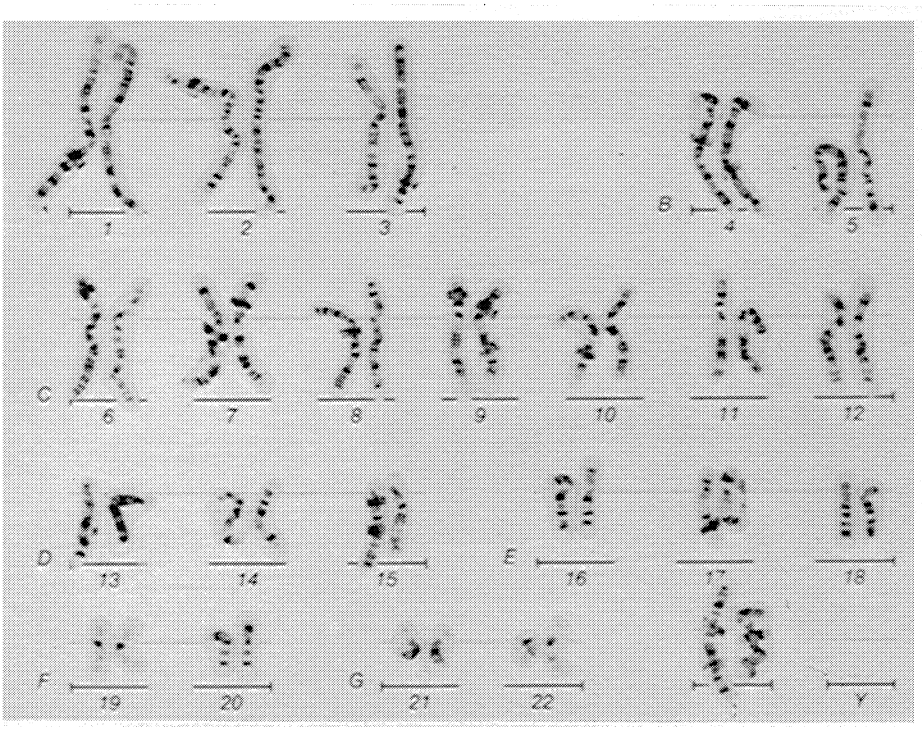

Figure 2. Normal female karotype.

Sex Determination

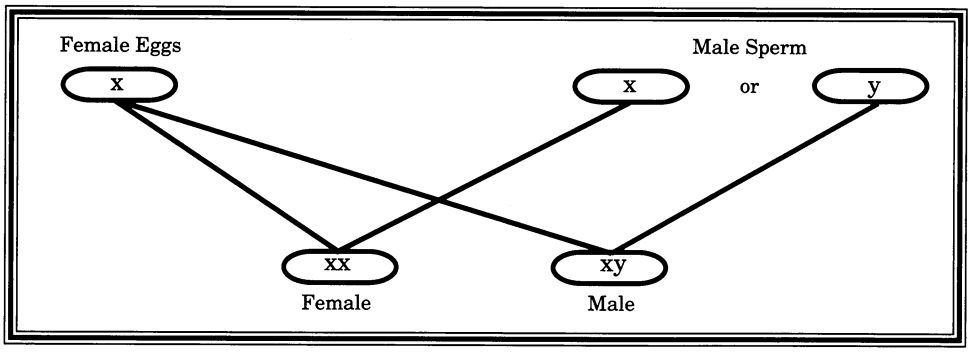

Figure 3. Sex Determination.