MEDICAL ASPECTS OF MENTAL RETARDATION

MEDICAL ASPECTS OF MENTAL RETARDATION

Compiled and Edited by

CHARLES H. CARTER, M.D., F.A.A.M.R.

Medical and Research Director
Sunland Hospital
Orlando, Florida

CHARLES C THOMAS · PUBLISHER
Springfield · Illinois · U.S.A.

Published and Distributed Throughout the World by
CHARLES C THOMAS • PUBLISHER
BANNERSTONE HOUSE
301-327 East Lawrence Avenue, Springfield, Illinois, U.S.A.
NATCHEZ PLANTATION HOUSE
735 North Atlantic Boulevard, Fort Lauderdale, Florida, U.S.A.

Library of Congress Catalog Card Number: 64-16090

With THOMAS BOOKS careful attention is given to all details of manufacturing and design. It is the Publisher's desire to present books that are satisfactory as to their physical qualities and artistic possibilities and appropriate for their particular use. THOMAS BOOKS will be true to those laws of quality that assure a good name and good will.

Printed in the United States of America
N-1

CONTRIBUTORS

STANLEY M. ARONSON, M.D.
Professor of Pathology
State University of New York
Downstate Medical Center
Neuropathologist, Institute of Pathology
Kings County Hospital Center
Brooklyn, New York

JAMES H. AUSTIN, M.D.
Associate Professor of Neurology
University of Oregon Medical School
Portland, Oregon

CLEMENS E. BENDA, M.D.
Director of Research and Psychiatry
Walter E. Fernald State School
Associate Professor in the Department of Psychology
Clark University
Worcester
Lecturer in Pediatrics
Tufts Medical School
Instructor at Harvard Medical School
Assistant Psychiatrist
Massachusetts General Hospital
Boston, Massachusetts

KINGSLEY BISHOP, M.D.
Director of Cyto-Biological Research
Sunland Hospital
Orlando, Florida

CECIL G. BUTT, M.D.
Pathologist and Director of Laboratories
Orange Memorial Hospital
Orlando, Florida

THOMAS BUTTERWORTH, A.B., M.D., Med. Sc.M.
Assistant Professor of Dermatology
Graduate School of Medicine
University of Pennsylvania
Chief, Department of Dermatology
Pennhurst State School
The Reading Hospital

Consultant in Dermatology
Berks County Tuberculosis Sanitorium
Pottstown Hospital
Wernersville State Hospital
Reading, Pennsylvania

C. H. CARTER, M.D., F.A.A.M.R.
Medical and Research Director
Sunland Hospital
Orlando, Florida

JAMES R. COOK, M.D.
Internal Medicine and Endocrinology
Orlando, Florida

WILLIAM JAMES CULLEY, Ph.D.
Director, Mental Retardation Research Laboratory
Muscatatuck State School
Butlerville, Indiana

ALFRED M. FREEDMAN, M.D.
Professor of Psychiatry and Chairman of the Department
New York Medical College
New York, New York

STERLING D. GARRARD, M.D., F.A.A.P.
Associate Professor of Pediatrics
State University of New York
Upstate Medical Center
Syracuse, New York

E. A. GIBBS, M.D.
Professor of Neurology
University of Illinois College of Medicine
Chicago, Illinois

FREDERIC A. GIBBS, M.D.
Professor of Neurology
University of Illinois School of Medicine
Chicago, Illinois

IRMA HAUSER, M.D. (RETIRED)
Pediatric Consultant with Women's General Hospital
Philadelphia
New York State Orthopedic Hospital
New York State Department of Health (Pediatrics)
Westchester County Department of Health (Pediatrics Division)
Private Practice

HAROLD E. HIMWICH, M.D.
Director, Research Division, Thudichum Psychiatric Research Laboratory
Galesburg State Research Hospital

Professorial Lecturer in Physiology
University of Illinois College of Medicine
Chicago, Illinois

WILLIAMINA A. HIMWICH, Ph.D.
Medical Research Associate IV
Galesburg State Research Hospital
Galesburg, Illinois

DAVID YI-YUNG HSIA, M.D.
Professor of Pediatrics
Northwestern University Medical School
Director, Genetic Clinic
Children's Memorial Hospital
Chicago, Illinois

ANDREW E. LORINCZ, Ph.B., B.S., M.D., F.A.A.P.
F.A.A.P. Associate Professor
Department of Pediatrics
University of Florida College of Medicine
Gainesville, Florida

JOHN H. MENKES, M.D.
Assistant Professor of Pediatrics
Johns Hopkins Hospital, Baltimore, Maryland
Joseph P. Kennedy, Jr. Memorial Foundation
Senior Research Scholar in Mental Retardation

JAMES H. McCLELLAND, Jr., M.D.
Superintendent
Polk State School and Hospital
Polk, Pennsylvania

A. S. NORRIS, M.D.
Associate Professor of Psychiatry
State University of Iowa
Iowa City, Iowa

WINTHROP M. PHELPS, M.D
Medical Director
Children's Rehabilitation Institute
Baltimore, Maryland
(Affiliated with The Johns Hopkins Hospital and Medical Institutions)

CHARLES M. POSER, M.D.
Professor of Neurology
University of Missouri School of Medicine
Kansas City General Hospital and Medical Center
Kansas City, Missouri

ISABELLE RAPIN, M.D.
Assistant Professor Neurology and Pediatrics
Albert Einstein College of Medicine
New York, New York

JULIUS B. RICHMOND, M.D., F.A.A.P.
Professor and Chairman
Department of Pediatrics
State University of New York
Upstate Medical Center
Syracuse, New York

IRA M. ROSENTHAL, M.D.
Associate Professor of Pediatrics
University of Illinois College of Medicine
Attending Pediatrician
University of Illinois Research and Educational Hospital
and Cook County Hospital
Chicago, Illinois

JACK H. RUBINSTEIN, M.D.
Director
Hamilton County Diagnostic Clinic for the Mentally Retarded
Cincinnati, Ohio

PHILIP SCHWARTZ, M.D.
Director, Dept. of Pathology
Warren State Hospital
Warren, Pennsylvania

FRANK W. SHAFFER, M.D
Associate Attending Pediatrician
St. Christopher's Hospital for Children
Associated in Pediatrics
Temple University
Montgomery County Developmental Center and School for Handicapped Children
Norristown, Pennsylvania

BRUNO W. VOLK, M.D.
Director, Isaac Albert Research Institute
Jewish Chronic Disease Hospital
Brooklyn, New York

EDGAR N. WEAVER, M.D.
Neurological Surgery
Roanoke, Virginia

MRS. ETHEL WILSON
Special Assistant to the Chairman
Department of Psychiatry
New York Medical College
New York, New York

INTRODUCTION

CLINICAL MENTAL RETARDATION

Definition

MENTAL RETARDATION or deficiency is a condition in which we find the individual's mental processes are below the accepted normal, due to arrested or incomplete mental development arising from genetic causes, or, induced by disease or injury before adolescence. When this condition develops after adolescence, it is more commonly known as Dementia.

Classification

1. Idiots	(Severely Retarded)	I.Q. 0-20
2. Imbecile	(Trainable)	I.Q. 20-49
3. Moron	(Educable)	I.Q. 50-70

In this classification, etiological factors are not considered. Each may further be classified into low, middle, and high. Emotional conditions alone can cause variations in I.Q. testing from 20 to 45 points.

From an etiological standpoint, mental retardation is divided into *two* large groups:

1. Primary or Endogenous
   Defect determined largely by causes inherent to the genetic constitution of the patient, i.e., multiple genes, single, dormant or recessive genes.
2. Secondary or Exogenous
   Caused by environmental factors: infections, toxins, endocrine pathology, trauma or metabolic imbalance.

A diagnosis of Mental Retardation must be recognized as a description of a symptom common to many different conditions, some clearly heritable and others in which inheritance has no part. Also, many reactions thought to signify mental retardation are due to physical discomfort or pain, especially in children six months

to three years of age. Mental alertness, responsiveness and intelligence may be suppressed in illness and even convalescence. Poor nutrition, environment, fever and even emotional conditions may depress mental activity and on continuation of such depression, mental retardation may become fixed.

Therefore, we must keep in mind that although it seems probable that a retarded mentality will be the outcome, there is the chance that damage has not been irreversible. And so, "if we of the medical profession," to quote Dr. Nichel, "are to keep these people alive, then we must see to it that they do more than breathe; their lives must at least be worthwhile to themselves."

PREFACE

PREPARATION OF a textbook dealing with the medical aspects of mental retardation is an extensive undertaking for one volume. Several books have been published recently which deal with certain divisions of the subject. This book resulted from repeated suggestions that there is need for a volume concerned entirely with medical knowledge and recent advances of interest to physicians, medical students and scientists interested in this problem.

Mental Retardation is perhaps the greatest single disabling condition afflicting the human race; the only satisfactory solution is prevention, and, the only means of prevention depends on better understanding of its cause. It is hoped that this publication will stimulate its readers by a better understanding of the multiple causes involved and is intended to bring many of the recent treatment advances into focus. It is also hoped much of the material in this book will soon be out-dated by a rapid, further advance of knowledge concerning the problem of mental retardation.

Much information herein contained is available in various publications. An attempt is made to bring this scattered data into one readily available volume. It would be impractical to cover every condition that could possibly be associated with mental retardation; however, the important facets must be discussed and may save a review of various books and the perusal of medical journals in order to find the desired information. This book is planned to aid the busy physician in diagnosing cases and in counseling parents of retarded children, thereby increasing mutual confidence.

The authors' intent is to discuss each subject concisely and to include rather extensive bibliography for those who are interested in further study. All contributors are persons particularly competent in each of the fields assigned. They write on the various subjects without censorship.

Mental retardation has been recognized throughout the history

of our civilization. At times, those afflicted, were regarded as persons to be destroyed or discarded and, in other cultures, as persons to be honored and almost worshipped since they were believed to be possessed by abnormal or powerful spirits.

Earliest medical references to mental retardation are probably found in Thebes Therapeutic Papyrus, 1552 B.C., and, was mainly classed as a form of witchcraft. In 1657, Wolfgang Hoefee brought forth the concept of air as a causative factor, and, in 1843, Little drew attention to neonatal brain damage as a cause. In 1861, Broca discussed local brain damage as a cause; social studies of the Kallihak family introduced heredity, and, about 1934, Folling directed attention to faulty metabolism.

Mental retardation occurs in approximately one out of every thousand births. However, only one per cent of these will be a complete burden on society; the other ninety-nine per cent under present care and treatment, will to varying degrees, be self-sustaining and contribute to society.

This condition has been defined in many ways. Possibly one of the best definitions is the inability of an individual to function at the mental level which has been accepted as an average for the general population. These are the persons who require special training and preparation in order to achieve the most in society.

Economic impact of mental retardation on society has been very great. Institutional care throughout the United States costs millions of dollars yearly; the loss of productivity of these persons represents a loss of many more millions. The social impact is very great in that many families are completely disrupted by the presence of an abnormal child in the family. Our divorce rate in marriages where there is a retarded child is over twice that seen in the population as a whole, and, the suicide rate in these families is almost three times higher. In the past, many of these children were considered mentally ill and kept in mental hospitals. Many were hidden from society. During the past twenty-five years, due to advancing public understanding, the handicapped child does not now carry the extreme social stigma it did formerly.

A gradual decrease of 25 per cent has occurred in the number of mentally retarded children during the last twenty-five years. Improved prenatal and postnatal care have contributed greatly

to this improvement. Congenital syphilis, injuries at birth, nutritional problems, parasitic infestations, infections in and around the central nervous system due to bacteria, have all been greatly decreased in number. Understanding of metabolism and its correction as in cases of phenylketonuria and galactosemia has also been important. Research, better diagnosis and treatment of these conditions must be advanced to the extent of our ability in order to speed further reduction of this unfortunate condition.

Mental retardation results from physical influences which impair normal development and function of the cells and structures in the central nervous system, particularly the brain. These changes occur either during prenatal or neonatal development, or, during delivery. There may be one or more causes such as fixed brain lesions, metabolic disorders, chronic somal lesions, endocrine disturbance, infections and various other and often obscure causes. Most of the severe disturbance of development occurs before the third month of pregnancy.

At twenty-two weeks, the brain is a homogenous, undifferentiated structure. At birth, differentiation is present but its development is not complete. There is absence of myelination of parts of the cortex; pyramidal tracts are not fully developed and coordination between various parts has not been established. Myelination of the optic nerves is not complete until eight to twelve weeks after birth. Most of the early neonatal intrauterine movements are entirely spinal, so we have practically a "State of Decerebration." Infants, who were found to have only a small, undeveloped cerebrum, were observed to breathe, cry, suck and have a grasp reflex. Maturation continues rapidly after birth until all parts are completely integrated. Although there are still many debated questions and details of histology and anatomy, attention is turning rapidly to the development of our knowledge of the biochemical processes which parallel the anatomical development, both normal and abnormal.

A study and understanding of the cellular chemistry and many changes occurring during the rapid development of the embryo are imperative for the appreciation of prenatal pediatrics, as well as the neonatal complex cellular activity.

We are becoming aware of an increasing variety of disorders

associated with a metabolic block and with recessive inheritance. They are of infrequent occurrence in private practice and it is important to recognize them early since prompt treatment may prevent permanent impairment and other pernicious effects. An enzyme defect may provoke mental disorder either through the accumulation of toxic metabolites in grossly abnormal amounts or through the absence of an end metabolite which is essential for normal development. If intracellular activity is altered, no cell can undergo either suspended or superactivity for long without showing irreversible changes.

An attempt is made herewith to classify conditions and diseases which bear on the clinical picture of mental retardation:

I Genetic factors, distorted or mutated genes.
II Metabolic disorders—proteins, carbohydrates and fats.
III Infections—parasite, virus, bacteria.
IV Injury—prenatal and postnatal.
V Disorders of the endocrines.
VI New growths—benign and cancerous

The brain doubles its weight during the first two years of life from approximately 400 grams at birth, to 1100 grams at one year, reaching its maximum by the end of the seventh to tenth year, then weighing about 1200 grams. Intellectual development continues throughout life.

A great deal of damage often can exist without modifying behavior in early infancy. Lesions of basal ganglia may be completely unnoticed until the more advanced activities of walking, talking, etc. begin. Spastic neck reflex in an infant or very young child should be considered with great concern as indicative of central damage. Brain cells cannot be regenerated or new cells formed, but, if destruction is not too extensive, new pathways may be formed to compensate.

The degree of mental retardation is in proportion to the time of onset, duration and severity of destructive factors, extent and location of the destruction. The development period at which slowing of growth rate occurs often determines the *type* of deformity and the *degree* of abnormality.

Editorial appreciation must be expressed to Dr. Eugene H. Payne, Dr. Andrew E. Lorincz, Dr. Howard V. Bair, Dr. Peter Bowman, Mrs. E. G. Blankenship, Mrs. E. E. Gillette, Mrs. L. P. Root and Mrs. J. R. Speicher for their untiring efforts on behalf of this publication.

We sincerely hope this book will contribute to the growing interest in this field.

C. H. C.

CONTENTS

MEDICAL ASPECTS OF MENTAL RETARDATION