American Handbook of Psychiatry

Advances in the Prevention and Treatment of Mental Retardation

Frank J. Menolascino
Fred D. Strider

Advances in the Prevention and Treatment of Mental Retardation

Frank J. Menolascino and Fred D. Strider

e-Book 2016 International Psychotherapy Institute

From *American Handbook of Psychiatry: Volume 7* edited by Silvano Arieti

Table of Contents

ADVANCES IN THE PREVENTION AND TREATMENT OF MENTAL RETARDATION

Frank J. Menolascino and Fred D. Strider

In this chapter current approaches to the prevention and treatment of mental retardation will be reviewed with a focus on specific psychiatric treatment and intervention appropriate to the combined syndromes of mental retardation and mental illness. In a concluding section, the authors will indicate their views regarding the essential elements of comprehensive approaches and treatment.

Introduction

The model of *primary* (prevention of the appearance of a disorder), *secondary* (very early diagnosis, effective treatment, and return of the person to a normative state), and *tertiary* (minimization of the remaining handicaps and return of the person to as high a level of functioning as possible) prevention will be utilized to review the currently available and possible future preventive approaches in each of these three dimensions. Although this three-step approach may seem simple, it is actually quite complex because there are over 350 causes of mental retardation. Major prevention programs have been successful in certain states: for example, Illinois has been successful in the area of screening and prevention of lead poisoning;

California has energetically encouraged public education concerning mental retardation; Connecticut has mounted an excellent program to prevent Rh blood incompatibility; and Massachusetts has a well-established program for discerning a number of preventable forms of inborn errors of metabolism. The interconnections between different levels of prevention and ongoing service delivery, and possible linkages between the different human service systems that serve the mentally retarded will also be discussed. This section will examine the continuity between prevention and clinical treatment of the physical and psychiatric symptoms of mental retardation.

Current Status of Prevention Efforts

A recent national report entitled "Preventing Mental Retardation: More Can Be Done" noted that, at the national level, the current "state of the art" in the prevention of mental retardation is fragmented and, while promising much, delivers little. There are always major lags between the evolution of new knowledge of mental retardation and its direct application in the field. The gap is currently being narrowed by advocacy, public information programs, and such exceptional programs as the regionalization and expansion of metabolic screening of newborns for the inborn errors of metabolism. However, federal funds have frequently been withdrawn from excellent pilot programs in prevention, and the states have not always been able to maintain enough programs even though these preventive efforts are

cost-effective and the lack of money “up front” results in increased costs and unnecessary human suffering later on.

In the United States, there are eight areas of activity in mental retardation in which preventive efforts are currently being implemented—albeit with some major gaps: (1) comprehensive prenatal care (including recent increased attention to “high risk” pregnancies and prenatal nutrition); (2) infectious diseases (both prenatal and postnatal); (3) chromosome disorders; (4) metabolic diseases (such as the inborn errors of metabolism); (5) internal (for example, Rh blood incompatibility) and external (for example, lead poisoning) intoxications; (6) adverse early childhood experiences within the family; (7) childhood accidents; and (8) other approaches such as screening for neural tube disorders. Each of these areas will be briefly reviewed.

Comprehensive Prenatal Care

The dearth of comprehensive prenatal services is a significant national problem. Low birth weight and prematurity are often associated with the symptoms of mental retardation, epilepsy, and cerebral palsy. A major issue in prenatal care is malnutrition during pregnancy, which may cause infant death or permanent brain impairment. Excessive maternal alcohol consumption or drug use raise the risk of having an abnormal infant to almost 44 percent.

After birth, the vulnerability of the developing brain to permanent damage as a result of inadequate nutrition has been clearly implicated. This susceptibility has been observed in both experimental animals and human beings. Nutritional supplement programs must include pregnant and lactating women, infants, and preschool children. Since the brain is still very rapidly developing during the first six years of life, one of the great advances in national health care in the last quarter century has resulted from improved general nutrition; this advancement must be vigorously extended to the poor.

National and state statistics indicate that many women still receive insufficient prenatal care, even though federal and state programs have been established to reach persons in economically depressed areas who might not otherwise receive such services. The extent of need for additional services is unknown because neither federal nor state agencies have adequately analyzed the extent of current prenatal care needs, the areas of greatest needs, or the effect of existing prenatal programs.

Following conception, comprehensive prenatal care can help prevent low birth weight and prematurity, which are in turn directly associated with mental retardation. This is the point at which early detection of and direct treatment intervention for chronic conditions (for example, diabetes mellitus, essential hypertension, hypothyroidism,) in the mother are critical. The identification of “high risk” pregnancy, in conjunction with the rapid initiation

of specialized obstetrical care, can save lives and minds, and can also save dollars in long-term care. The Vermont Association for Retarded Citizens recently completed a study that concluded that a comprehensive perinatal program should be composed of the elements (noted in figure 31-1) that delineate the mechanics of dealing with “at-risk” pregnancies.

The components indicated by this flow chart are neither new nor are they expensive to initiate. This approach encompasses putting together the already known basic components of a successful prenatal program. As to linkages, the Vermont plan for perinatal care recommended that the perinatal program be regionally based throughout the state via a strong university and department of health collaboration. Although Vermont is a small state, it is interesting that it felt that its projected perinatal program could build on current resources by adding—over a four-year period—the following components to bring the program to fruition: (1) family planning and counseling services; (2) genetic screening and counseling; (3) maternal/fetal transportation; (4) high-risk obstetrical unit; (5) intensive care of the newborn; and (6) program evaluation

A greater level of awareness by family physicians and family planning centers of the importance of high-risk pregnancy identification is needed. A key factor is the availability of the mother’s obstetrical history, which can yield the necessary information for a prompt referral to regional diagnostic

programs for the further detection of, and direct dealing with, high-risk pregnancies. Increased funding and resources will be needed to carry out these prenatal efforts in our country, and the system (or individual programs) to be utilized can be a series of specialized units in regional general hospitals or a medical service system that is integrated into current generic health services.

TEENAGE PREGNANCIES

The alarming number (20 percent of all live births and 26 percent of low-birth-weight infants) of teenage pregnancies directly contributes to increased numbers of premature infants and infants who are gravely "at risk" to develop mental retardation. Adolescent pregnancies increase health risks, such as higher incidences of toxemia, anemia, prolonged labor, and, for the infant, increased incidences of low birth weight and related signs and symptoms of mental retardation.

Teenage pregnancy is a pressing social problem compounded by ignorance, immaturity, illness, illegitimacy, and poverty. It costs billions of dollars each year in welfare support and medical costs. It has been estimated that about half of the women currently on the welfare rolls had their first child during adolescence. A significant number of teenagers are not adequately informed about, or disregard, contraception. Every year, more

than one million teenage girls, most of them single, become pregnant; many are younger than fifteen. These pregnancies result in about 600,000 live births a year. The others are terminated by miscarriages (100,000) and abortions (300,000). Teenage mothers give birth to a disproportionate number of premature babies born with low birth weight (under 2,500 grams): 16.5 percent for women under age fifteen; 10.1 percent for women age fifteen to nineteen. Among very small premature babies (under 1,500 grams), the incidence of mental retardation in those who survive is about 26.3 percent compared to only 1.6 percent for full-term babies.

Teenage motherhood brings many complications. Many young girls are ill informed of medical care needs and do not receive adequate prenatal care or nutrition. Others, especially those in their early teens, ages eleven to fourteen, are not physically mature and are often unable to bear the stress of having a baby without high risk to mother and child.

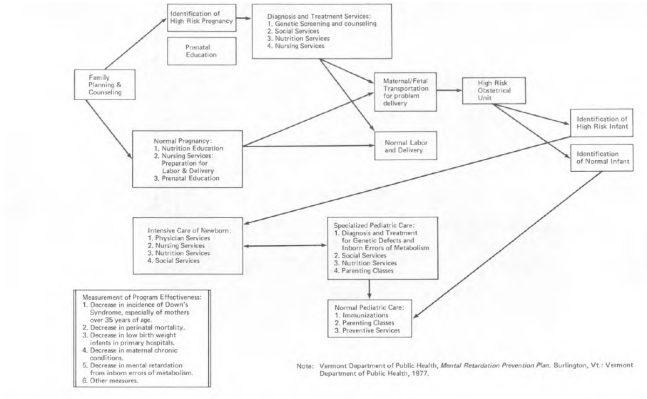

Figure 31.1.
Perinatal Program Flow Chart

Beyond the medical concerns related to adolescent pregnancies are some difficult social issues: (1) the low priority of the adolescent in our society; (2) the conspiracy of silence and lack of accurate information regarding human sexuality; (3) lack of access to medical services, including contraceptives; and (4) the influence of a society with a negative view of sex.

Currently, our country's health services delivery systems do not provide one-stop centers offering comprehensive pregnancy-related services, including counseling oriented to the prevention of teenage pregnancy. Also not provided are the mechanisms for extended follow-up necessary to provide needed services to pregnant teenagers whose offspring may be "at risk" for mental retardation. However, a bill enacted by the Ninety-fifth Congress (S. 910 by Kennedy, Williams, Javits, and Hathaway) has established

federal grant support for networks of community-based services to prevent initial and repeat pregnancies among adolescents. Further, there is congressional action pending to amend the Maternal and Child Health Act (Title V of the Social Security Act) and other pertinent legislation (such as Title XIX of the Social Security Act) to establish a network of "risk centers" for women whose pregnancy presents high risk, with special emphasis on the problems of teenage pregnancy. These congressional actions can well result in the initiation of a series of pregnancy "at-risk centers" throughout our country. These centers will require a close liaison (and subsequent linkages) between obstetric, pediatric, mental health, visiting nurse, and social work resources in order to be most effective.

Infection

Infection, both before and after birth, has been a continuing cause of mental retardation. Prior to 1963, the prevention of these infections was viewed as an improbable development. In that year a vaccine became available for measles and, in 1965, a vaccine for rubella was produced. The incidence of mental retardation resulting from measles and rubella has plummeted, but the immunity levels in the general public are still unnecessarily low; death and residual symptoms from these infections still occur.

The topic of infectious diseases, which affect both the unborn and the born, still requires more research attention. For example, meningitis strikes over 20,000 citizens each year and its cardinal signs and symptoms should be made common knowledge in order to increase detection of the disease very early in its course. Few field studies of available vaccines for the most frequently noted bacterial agents that produce meningitis have been made. However, the increasing demand (and establishment) of mandatory immunization of children before school entrance is a nationwide trend.

A recent General Accounting Office (GAO) Report pointed out that immunity levels in the general population have not been adequately assessed to identify areas in which immunization levels are low, nor have vaccination programs raised immunity to acceptable levels. The National Center for Disease Control still considers immunity levels to be too low in our country. For example, it is estimated that over 10 percent of all pregnant women are at-risk of contracting rubella. This is a sad commentary when a blood sample from the pregnant woman can be analyzed to determine whether she is immune to rubella or requires effective treatment.

As of September 1977, forty-seven states had laws requiring rubella immunization before school entry. Though the Headstart program guidelines require that each enrollee have a physical examination, including screening for immunization status, this has been waived in some states and so only

incomplete records are available. From a national perspective, two elements are necessary to effectively combat these diseases: (1) comprehensive data on immunity levels in the child and adult female population to pinpoint problem areas; and (2) aggressive immunization programs targeted on areas with low immunity levels. Utilizing these guidelines, at least 95 percent of all children would be immunized before they entered school and rubella titers would be a part of all family planning services. Private physicians must be educated to be highly conscious of the medical-legal implications of providing this protection to women of childbearing age.

As with preschoolers' immunizations, rubella titer evaluation and follow-up immunization before entering high school should be made mandatory. Since vaccination must precede conception by two months, at the time of marriage a reevaluation should be required and a vaccination or booster given if the antibody titer is not adequate. This vital preventive step could be legislated by the federal government as a prerequisite for obtaining a marriage license.

Certain federal programs, in particular Headstart and family planning programs, can improve surveillance data and possibly raise immunity levels. These programs are all important linkages to other components of a state's approach to prevention. Mandatory vaccination, before entering school, against diphtheria, pertussis, tetanus, measles, rubella, and poliomyelitis is a

current trend that has been firmly supported by recent federal government statements. The Department of Health, Education and Welfare (HEW) committed an additional 3 million dollars for childhood immunization programs in 1979; and former HEW Secretary Califano made mandatory immunizations an integral part of the Child Health Assessment Program, which focuses on the health status of young children.

The normal childhood diseases (mumps, measles, chicken pox) can occasionally result in encephalitis with subsequent mental retardation as a residual. Supportive treatment of these "normal" childhood diseases, which infrequently have "non-normal" outcomes, must be actively sought.

Chromosome Disorder

Chromosome abnormalities are estimated to account for about 16 percent of institutionalized mentally retarded citizens. Amniocentesis makes possible the prenatal diagnosis of an increasing number of developmental disorders such as: chromosome abnormalities (Down's syndrome), neural tube defects (meningomyelocele, anencephaly), inborn errors of metabolism, and sex-linked genetic disorders. The need for an amniocentesis evaluation is appropriate when the following factors are present: increased maternal age (over thirty-five years), maternal carrier of X-linked diseases, previous trisomic offspring, previous neural tube defect, maternal carrier of an

established chromosome translocation, parental carriers of inborn errors of metabolism, and high-risk pregnancies with repeated miscarriages. Amniocentesis may also be indicated when there is distinct evidence of genetically-based instances of severe mental retardation in the family. An obstetrician and/or genetic counselor may recommend that amniocentesis be performed between the fifteenth and eighteenth weeks of pregnancy. About 5 percent of mid-term pregnancies (250,000 yearly) are considered "high risk" and would benefit from the amniocentesis procedure.

Contrary to some current opinions, many children's lives are saved by genetic counseling and amniocentesis evaluation. For example, if the amniocentesis procedure showed the presence of a normal child, then recourse to abortion is unnecessary. If a Down's syndrome infant is detected by amniocentesis, then the parents can make specific plans for early physical intervention, psychosocial stimulation early in life, and similar treatment-management approaches that can serve to ameliorate the developmental delay. Continuing and increased availability of amniocentesis, in the authors' opinion, will also accelerate the development of methods for very early treatment *in utero* that may ameliorate any prenatally diagnosed conditions.

Currently, genetic screening and counseling services are only readily available to the higher income groups, while the majority of people who would benefit from such services are not receiving them. Also, an insufficient

number of at-risk mothers are not receiving these services because the needed pool of trained manpower does not exist. However, current research in selectively detecting fetal cells that have intermingled with the maternal bloodstream during early pregnancy, holds promise for replacing the current amniocentesis technique with a relatively simple peripheral blood sampling test—thus enhancing the availability of prenatal diagnostic services.

The incidence of neural tube disorders is related to genetic factors and the need to screen for these disorders. Yet screening for neural tube disorders (that is, alpha-fetoprotein levels in peripheral blood and amniotic fluid, via amniocentesis) should become a routine aspect of all prenatal care programs.

A concise review of the indicators for chromosomal disorders in pregnant women should be routinely utilized in prenatal care in all health programs. Outreach into the lower socioeconomic groups is needed. Because family planning programs contact so many women, these programs are ideal as sites for outreach activity. The referral consciousness of the family practitioner in regard to potential chromosome disorders in at-risk mothers must also be raised.

Metabolic Disorders

Metabolic disorders and chromosome abnormalities are separated because the nature of the diseases and methods of diagnosis are, at times,

very different. Many hereditary metabolic disorders can be diagnosed in the newborn and a few (Methylmalonic acidemia) have been detected and treated *in utero.* The classical illustration of the detection of an inborn error of metabolism in the newborn period is phenylketonuria (PKU). Disorders such as hypothyroidism (twice as common as PKU) are also detected by means of newborn screening, and treatment is available.

A number of model state laws have appeared in the last two decades mandating the screening of newborn infants for metabolic disorders that are associated with mental retardation. These screening techniques have been automated to make them more cost-efficient. Expansion of metabolic screening to include many other treatable disorders is feasible; with automation the cost of multiple screening may be little more than that of one routine PKU test. For example, the following disorders can be detected early and rapidly by mass screening: (1) PKU; (2) hypothyroidism; (3) Galactosaemia; (4) valinemia; (5) homocystinuria; and (6) histidinemia.

Yet, much remains to be accomplished. A national survey of newborn screening to which forty-three states responded, noted: Six states were not conducting PKU screening programs; thirteen states were screening for Galactosaemia; seventeen states were screening for hypothyroidism; and fifteen states were carrying out additional tests, some of which included alanine deaminase deficiency, histidinemia, lead poisoning, sickle cell disease,

thalassemia, leucine deficiency, (such as, maple syrup urine disease), methionine deficiency, Tay-Sachs disease, and toxoplasmosis. In the twenty-one state questionnaire returns that were usable, a composite 95 percent coverage for PKU was found. Two states have programs to detect maternal phenylketonuria early in pregnancy so that a reduced phenylalanine level diet can be instituted to protect the baby *in utero.* Screening requirements are, to some extent, specific for different populations, and transient neonatal phenomena are responsible for many abnormal results. It is important to have sound methods for confirmation and follow-up.

PKU, which occurs in one in 10,000 newborns, has become the classic model for illustrating the prevention of mental retardation. In the past, 1 percent of the incidence of severe retardation was secondary to PKU. Now these tragic consequences can be avoided by a phenylalanine controlled diet.

Six states attempt to detect maternal PKU by assessing elevated phenylalanine blood levels present in the neonate. Maternal phenylketonuria is an emerging problem, since the symptoms of mental retardation are a consequence of the infant being literally saturated *in utero* by high concentrations of phenylalanine in the mother's bloodstream. Routine screening of expectant mothers for PKU should be followed by treatment of the maternal carriers with a reduced phenylalanine diet. The cost-benefit ratio for prevention of PKU is clearly in society's favor.

Newborn screening for thyroid deficiency has been actively pursued in recent years. Since the incidence of congenital hypothyroidism has been reported to be from one in 3,000 to one in 7,000 live births, it is clear that this disorder is a frequent metabolic cause of mental retardation. The clinical diagnosis of hypothyroidism in an infant is difficult, and the screening technique becomes both a crucial diagnostic and preventive tool. The initial mass screening is accomplished by a filter paper technique (the thyroid-4-component), and the positive samples are validated by further testing (the thyroid stimulating hormone component). This procedure has recently become operational in Michigan where the State Health Department coordinates both the screening (by young volunteers from the Illinois Association for Retarded Citizens) and treatment coordination (by the medical staff of general hospitals throughout the state). Their program is an excellent example of putting current knowledge about the prevention of mental retardation into direct service for young citizens. The condition of these infants would otherwise probably not have been detected within the first three months—the critical period of intervention. If treated with exogenous thyroid hormone within the first three months, the majority of the afflicted infants will develop normally.

Some maintain that "meddlesome" intervention with our genetic endowment, by permitting genetically abnormal people to survive and to reproduce, will be detrimental to our "national genetic balance" (that is, the

gene pool). However, it has been estimated that by allowing PKU patients to reproduce it would require over 100 generations for the gene frequency to double. It is only since the infant mortality rates started to fall that we have been able to differentiate more clearly genetic diseases from acquired diseases. The process of human evolution has been altered far more rapidly by antibiotics than by the advent of PKU or hypothyroid testing.

With automation of the assessment of direct blood specimens (via the punch-index machine), newborn screening tests for treatable inborn metabolic error diseases such as homocystinuria, tyrosinemia, Galactosaemia, maple syrup urine disease, histidinemia, and valinemia can be added to PKU testing without excessive addition of laboratory personnel. If the laboratory processes a minimum of 25,000 newborn specimens a year, the cost increase over PKU screening alone is minimal. It is most frequently recommended that a regional program be based in a university department of pediatrics in close alliance with the state department of health. This linkage is an essential one if the operational problems noted in the recent past in attempting to implement neonatal screening programs are to be avoided. Guthrie noted that for most states, the legacy of the 1960's controversy concerning PKU remained in the 1970's in the form of three problems: (1) lack of liaison between the medical centers and the screening programs; (2) many states with laws requiring PKU testing simply do not have sufficient population to make multiple testing of newborn infants practical; and (3) many states with large populations allow

each private laboratory to perform small numbers of PKU tests for a profit, thus causing the same problem that exists in small states. In May 1975, a regional model of linkage, the Oregon Neonatal Metabolic Screening Program, was established in Portland. It receives screening test specimens from Oregon, Alaska, and Montana. In January of 1976 the Massachusetts Metabolic Disorders Detection Program in Boston started receiving screening test specimens from Massachusetts, Rhode Island, and Maine.

Although an increasing number of the inborn errors of metabolism can be tested on a regional basis and the cost is only about $2.50 per complete testing, the follow-up of high-risk infants should be assured. This follow-up care should be available at special treatment-management regional centers (for example, local crippled children facilities, medical centers, or in conjunction with regional treatment centers) such as those established in California (California Assembly Bill #45, 1977). Experiences in England, Sweden, and France have clearly documented that over 50 percent of the motor and special sensory handicaps noted in developmentally at-risk newborn children can be completely reversed, to the great developmental enhancement of these children. Accordingly, early secondary prevention efforts can inadvertently become tertiary preventive challenges if follow-up services are not available after early diagnosis has been accomplished.

Intoxications

Lead Poisoning

Lead intoxication and Rh blood incompatibility disease are fine examples of primary preventive accomplishments in the field of mental retardation. Recognition of childhood lead poisoning is relatively new—it was first described in children in the early part of the twentieth century. The cause was assumed to be the ingestion of paint, but the role of air-borne lead in general exposure—primarily from automobile emissions—has recently received increasing attention.

There has been a clear recognition of lead's ubiquity, and intervention has taken place on the federal legislative level with the "Lead Paint Act," and planned reductions in the content of lead in gasoline. Young children up to the age of six years are at great risk, especially from peeling paint and chipping plaster in dilapidated housing. There is a strong association between pica and lead poisoning. Programs have been instituted to eliminate asymptomatic and symptomatic lead poisoning. The free erythrocyte protoprophyrin (FEP) test, which can be conducted from a drop of blood on filter paper, has made wide-scale testing feasible. Spinoffs of the FEP screening test include detection of iron deficiency anemia and the hemoglobinopathies.

Lead poisoning is not just an inner-city problem, but a widespread one that justifies more than a high-risk target area approach. This insidious and cumulative metal affects American children in epidemic proportions; over

600,000 bear blood levels above 40 mg/100 cc of blood. In 1970, the U.S. Surgeon General recommended that 40 mg/100 cc blood be the borderline at which the child is in potential danger of clinical lead poisoning; in 1975 the revised borderline was lowered to 30 mg/100 cc of blood. Ages one through six years of life are at the greatest risk for lead poisoning, with the peak years being from one to three. Theoretically, even very low levels of lead are associated with toxicity, and lead is known to cross the placenta during pregnancy.

Does a threshold for lead toxicity exist? On both theoretical and empirical grounds, there are several reasons to believe that if there is a threshold, many children have exceeded it. First, there is no known metabolic function for lead. Secondly, global environmental levels of lead have increased since the industrial revolution—most markedly since the invention of the automobile—and, in the evolutionary scale of time, the human organism has had only a brief opportunity to adapt to contemporary environmental levels. Third, no toxicologist would accept a margin of safety of 50 percent, yet thousands of children carry blood-lead levels of 40μg/dl. Lastly, the symptoms of lead toxicity are vague and easily missed. Headache, lethargy, colic, or clumsiness are not readily identifiable as symptoms of lead poisoning. Perino and Ernhart reported that black preschoolers with blood-lead levels above 50 μg/dl had significant impairment on the McCarthy Scales of Mental Development when compared to children matched for race and

controlled for other variables with blood-lead levels below 30 μg/dl. Undoubtedly, many children with these symptoms are misdiagnosed by both parents and physicians.

Of considerable interest is the report of Beattie and coworkers. They identified seventy-seven children with mental retardation of unknown cause and matched them with normal children on age, socioeconomic status, and geographic residence. The place of residence of the mother was identified and a sample of the drinking water was analyzed for lead. No normal children came from homes with excess lead, while eleven mothers of retarded children lived in homes with high levels of lead in the water during the time they carried the child. The authors concluded that the risk of retardation is increased by a factor of seven by living in a home which has a high lead level in the water during the mother's pregnancy.

Mass screening for lead poisoning utilizes the free erythrocyte protoporphyrin test (FEP), which consists of a dried blood sample collected on filter paper. Volunteers can be quickly trained to collect the dried blood specimens, and the samples are forwarded to a state (or regional) laboratory. The Illinois Association for Retarded Citizens has been particularly effective in organizing volunteers to screen large regions of the state of Illinois. Their testing technique (noted previously) and the more recent zinc-protoporphyrin test are inexpensive. Positive tests are further evaluated via

blood level determinations. The family physician carries out treatment with British Anti-Lewisite Factor (BAL) when necessary.

An effective program against lead poisoning must concern itself with a number of issues. Parents and the general public must be educated as to sources of lead, its effects, and what can be done. Screening with an inexpensive test (such as the free erythrocyte protoporphyrin test) is necessary because signs and symptoms of toxicity are too variable and covert. Following detection at the screening level, confirmation, and the institution of therapy, the child should be periodically monitored as to the effectiveness of the treatment—this may sometimes be necessary for up to two years. Finally, the environment of the child must be corrected to prevent re-exposure.

In summary, lead poisoning is directly linked to mental retardation, epilepsy, and cerebral palsy; it is a widespread problem, is frequently asymptomatic, and often presents with generalized or vague symptoms. The new free erythrocyte protoporphyrin test has made widespread screening for lead poisoning feasible. Although the total elimination of lead from our environment is probably not possible as a relatively short-term goal, the elimination of leaded gasoline is nearing reality.

Rh Incompatibility Disease

Hemolytic disease of newborns secondary to Rh incompatibility, is now

a preventable disease—only ten years after the introduction of routine post-birth administration of hyperimmune globulin (i.e., RhoGAM) to Rh-negative mothers. Careful adherence to clinical and preventive guidelines can help ensure that every susceptible woman is treated, so that the disease will be eliminated.

The latest available information (1974) reveals that there are over 7,000 infants born each year in our country who have sequellae of Rh-blood type incompatibility. Yet, only five states have Rh-blood type registries or education of the general public and professionals about the use of RhoGAM for preventing this disorder. An example of an excellent state-wide educational program on the use of RhoGAM is that carried out in Connecticut.

Immunoglobulin (RhoGAM) was licensed for human treatment in 1968. Although it can protect children from Rh incompatibility, it is currently underutilized. Problems exist because of its lack of availability in rural areas and failure to administer the agent to women after abortions as well as after births. Public and professional awareness must be raised concerning its vital preventive role. Prevention of Rh incompatibility includes the following issues: (1) early detection of all Rh-negative mothers; (2) availability of this information during pregnancy and delivery; and (3) prompt utilization of immunoglobulin after the delivery of each Rh-positive child to an Rh-negative mother. Yet, there is a lack of complete national information on the incidence

of Rh-negative and the accompanying immunization by immunoglobulin. For example, the recent GAO Report noted that only five states had mechanisms for fully monitoring Rh hemolytic disease (Connecticut, California, Illinois, New Jersey, and Colorado); only seven states required by law either premarital or prenatal blood typing; and only six states had special programs for immunoglobulin utilization, as of the date of the report. One would think that the private physicians would routinely check for Rh incompatibility, but the current Rh incidence rate indicates that this is not true. The consequences of not checking for Rh-negative incompatibility might lead to tragic consequences for the afflicted child.

Interestingly, Connecticut has had a complete Rh prevention program in operation for the last ten years. A recent report from the Center for Disease Control shows that Connecticut has had a 96 percent decline since 1970 in maternal sensitization and Rh blood disease in the newborn. Their prevention program includes routine premarital or prenatal blood typing. Comprehensive data are compiled on immunoglobulin usage at the time of birth, abortion, or miscarriage; Rh incidence and mortality (fetal and infant); the number of sensitized women; and the overall effectiveness of each of these preventive efforts are regularly monitored. State law in Connecticut requires that the Rh typing be listed on the marriage licenses, and each hospital is mandated to report information concerning Rh determination, the use of immunoglobulin, and follow-up results.

Mercury Poisoning

The hazardous situation occurring through large amounts of mercury being discharged into waterways was first recognized in 1970 when eating fish that had been contaminated was established as a definite link to human toxicity. There are two types of mercury poisoning, acute and chronic. The acute situation arises from the ingestion of soluble mercuric salts (mercuric chloride) and produces serious renal tubular damage. The chronic form develops from inhalation of mercury vapor or ingestion of small quantities of mercuric nitrate or other salts; it clinically presents with psychiatric and gastrointestinal symptoms.

At an increasing rate, alkyl mercury compounds have become significant environmental problems. The compounds ethyl and methyl mercury are soluble in organic solvents, and the covalent carbon/mercury bond is not biologically degradable. Intestinal absorption is nearly complete, and it is difficult for the body to rid itself of these compounds because of the continuous interchange of the gastrointestinal-liver blood circulation. Seed grain which has been treated with alkyl mercury compounds (such as antifungal agents) has been diverted into food, resulting in epidemics in Japan and several other countries. Methyl mercury is especially found in tuna and swordfish taken from waters contaminated with mercury. In humans, this compound freely passes the placental barrier, accumulating in the fetus, with

resultant cerebral palsy and mental retardation in the newborn.

It has been found that n-acetyl-DO-penicillamine, in a dose of 500 mg four times a day, accelerates the excretion of mercury and has been used successfully in the therapy of chronic mercury poisoning. However, this treatment is not effective in methyl mercury poisoning, in which case the use of ion-absorbing resin to interrupt the body circulation of toxic levels of mercury shows promise as a method of treatment.

The prevention of mercury poisoning will necessitate the shift of industries away from mercuric chloride, calomel, and mercury ointments—for all of which safe substitutes are available. This would eliminate acute poisoning and many instances of chronic poisoning. This shift to alternative compounds has already occurred to some degree. Mercuric nitrate has been eliminated from the felt-processing industry. Mercury salts for fingerprinting by police departments have been replaced by barium, zinc, or bismuth salts. Silver has replaced mercury in the manufactory of mirrors. This shift to alternatives, however, should be accelerated.

Early Childhood Experiences

The noxious developmental effects of adverse childhood experiences have, unfortunately, only recently been directly addressed by our society. Child abuse (physical, sexual, and emotional), secondary effects of severe

childhood mental illness (childhood depression and the psychoses), minimal opportunities for developmental stimulation secondary to poor parental modeling, racism, and the "culture of poverty" all represent major current prevention frontiers. Children born and reared in urban ghettos or impoverished rural areas are fifteen times more likely to be diagnosed mentally retarded than children from middle-class, suburban environments.

The Milwaukee Project was a research undertaking in which a target group of women from socioeconomically blighted census tracts in the metropolitan area of Milwaukee, Wisconsin, were studied. A very high frequency of mental retardation had previously been clearly documented in these census tracts. The research focused on two major approaches: (1) intensive training programs for pregnant mothers (for example, counseling and vocational training, instruction in mothering, child care, and homemaking); and (2) direct assistance to the families in stimulating their child by providing enriched early developmental experiences. The results clearly showed that the study group of children born to these mothers developed intelligence quotients averaging 25 points higher than the control group of children who were not provided these mother-child special training /stimulation experiences. This study indicates what can be done to alter socio-cultural causes of mental retardation by intensive environmental stimulation programs.

Unfortunately, most of the research in this area has not utilized the rigorous methodology employed in the Milwaukee Project. Indeed, definitive evaluations of the bulk of current environmental enrichment programs—and the people they are reaching— still needs to be determined. For example, the GAO Report notes that the University of Connecticut completed a national survey to determine what developmentally enriching programs for children under three years of age were operating or proposed. Information was solicited from several sources, including the state departments of education, state offices of child development, and early education program directors. A total of fifty-three ongoing and proposed programs were identified as operating at 116 sites and involving about 19,000 children and their families. Additionally, eight universities and twenty-three community colleges were involved in infant and toddler research and service programs. No clear set of program guidelines (or results) was discernible. The availability of these development enrichment programs is far below the demonstrated need for them. The national-state evaluation picture has changed little since this 1974 study.

The prevention of psychosocial retardation requires education, improved childrearing practices, and environmental correction for developmental high-risk infants. Ideally, programs would start early and continue until the child enters the formal school system, concentrating on definitive developmental corrections and on increasing verbal-social skills.

Attitudinal changes in the homes (and in school) will probably have the most long-lasting effect, since the intellectual/ educational atmosphere of the home is an important determinant of future development. Regional and state programs must work aggressively to train mothers of at-risk children (for example, low-income adolescent mothers). Unfortunately, it is very difficult to clearly delineate the linkages necessary in order to accomplish this goal. The complexity of this issue which literally cuts across virtually all areas of the concept of prevention is noted in table 31-1.

Accordingly, it appears that improvements in national policies, commitments to enhanced living standards, and increased human service provision can result in a Gestalten that acts favorably on the future lives of these psychosocially at-risk individuals. It is not the authors' intention to belittle the efforts of specific prevention programs, which can appreciably affect the noted key issues (for example, child abuse prevention projects), but to stress that the scope of this prevention challenge is a massive one since it eventually addresses key issues such as the persistence of "illiteracy."

TABLE 31-1 Relationship between Poverty and School Failure

I. Inadequate Intrauterine Environment due to:

A. Poor maternal growth

B. Poor maternal nutrition

C. Poor maternal health and health care

D. Too many pregnancies too close together

E. Extremes of maternal age (too young or too old)

F. Poor obstetrical care

II. Poverty:

A. Increased infant mortality

B. Increased infant morbidity

C. Elevated family size

D. Poor nutrition

E. Increased illness

F. Absence of adequate health care

G. Inadequate physical surroundings

H. Inadequate home learning environment

I. Inadequate home emotional environment

J. High level of social stress

III. Effects in childhood and adulthood

A. Increased chance of school failures and under-achievement

B. Adult unemployment and underemployment

Source: Birch, H.G., and Gussow, J.D. Disadvantaged Children: Health, Nutrition, and School Failure. New York: Grune & Stratton, 1970, p. 268.

Childhood Accidents and Postnatal Injuries

Childhood accidents and allied postnatal instances of head injury continue to be a major cause of mental retardation. Similarly, accidental

poisoning also remains difficult to prevent.

Accidents are the number one cause of death among infants and children. Although postnatal head injury seldom results in mental retardation, there is an increased incidence of residual behavioral disturbances and neurological handicaps. A recent Canadian report on this topic noted that preschoolers, with their energy and lack of judgment, are at greatest risk. Other predisposing factors are hyperactivity, aggressiveness, hunger, and tiredness. Accidents that can result in severe injury to the brain include auto accidents, falls, sports injuries, near suffocation or drowning, and extensive body burns. The American Academy of Pediatrics recently stated that approximately 90 percent of these childhood accidents are preventable.

Prevention strategies must include an increased public education program, promoting safety in the home and in sports, packaging of drugs in child-proof containers, and special hazards protection, such as auto seat restraints. Similarly, there should be stringent enforcement of child abuse and "driving while intoxicated" laws. There also needs to be an increase of correct information for parents about the signs and symptoms in a child that require immediate medical attention. The utilization of information services (for example, poison control centers) and similar public information can help in dispersing this information.

Other Disorders

There are many disorders for which the causes are not always known, but the disorders themselves can be ameliorated or prevented. For example, the role of nutritional supplements, both during pregnancy and in early life, has been shown to improve the physical integrity of infants. Accordingly, states such as California have passed legislation that automatically provides these supplements to low-income pregnant mothers, their infants, and their young children. This prevention program emphasizes the importance of giving all children an optimal chance for full physical development—even though the specific role of early nutritional intervention is heatedly debated among scientists.

Similarly, even though the exact causes of hydrocephaly and spinal cord disorders such as meningomyelocele (herniation of the spinal cord through its overlying bone and skin covering in the lower lumbar back region) are not known, therapeutic alternatives are available. Specifically, mothers who have had a previous child afflicted with one of these disorders can now be screened during subsequent pregnancies via a peripheral blood test for the alpha-fetoprotein (a protein that leaks out of the spinal fluid system of the fetus into the surrounding amniotic fluid, and hence into the maternal circulation). If the test is positive, it can be confirmed via amniocentesis, and the parents may be given the option of a therapeutic abortion. Interestingly, Dr. Robert Cooke has

recently suggested that abortion may not be necessary if we further explore the evolving research work on fetal surgery *(in utero)* on monkeys. This work provides a direct parallel for future interventions in humans—interventions that would both avoid abortion and provide the future infant with an intact central nervous system. The current hope for prevention of these disorders is best provided by the previously mentioned regional chromosomal screening/genetic study centers. Last, an enhanced awareness of this prenatal screening test and a rapid increase in genetic counselors will be needed to offer this alpha-fetoprotein assessment to all potential cases.

Priorities and Linkages

The prevention of mental retardation will have its greatest impact if attention is focused on the following areas: (1) optimizing preconception conditions; (2) improving pregnancy outcomes; and (3) optimizing infant growth and development. The activities necessary in each of these three areas constitute an intervention strategy for the prevention of mental retardation. This involves a Cycle of Continuing Intervention (see figure 31-2), which indicates that persistent and broad-based interventions are necessary to prevent mental retardation in future generations.

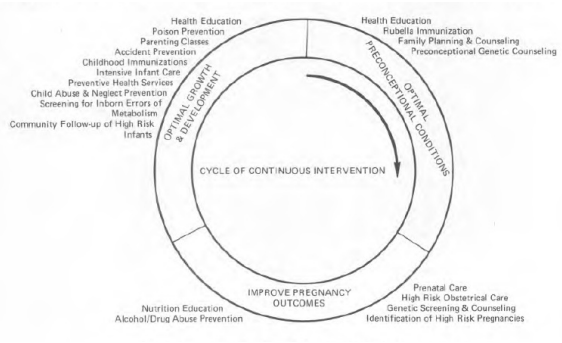

Figure 31-2.
Cycle of Continuous Intervention.
NOTE: Vermont Department of Public Health. Mental Retardation Prevention Plan, Burlington, Vt.: Vermont Department of Public Health, 1977.

A fully operational program for the prevention of mental retardation would contain all of the elements listed in the "Cycle of Continuing Intervention." Since the availability of financial resources may preclude the unilateral implementation of the prevention program, priorities should be established. One criterion for establishing priorities may be to choose those program elements that will have an immediate impact upon the prevention of mental retardation. Another criterion would be to choose those program elements that produce the greatest impact for the dollars expended. Together, these criteria indicate that the perinatal program be selected as a high priority item because of its ability to influence a number of factors that cause mental retardation. For example, the activities of a comprehensive perinatal program should produce a decrease in the number of low birth weight infants, chronic

maternal conditions, and genetically determined abnormal infants. The elements of such a comprehensive perinatal program were previously reviewed in the “Perinatal Program Flow Chart” (figure 31-1). In addition to indicating the necessary program elements, this chart also depicts the flow of a prospective patient through such a perinatal program.

Some of the perinatal program elements previously described are currently in place and operating in some of our states while other elements need to be developed or expanded. The following items are additional resource requirements that may be needed to produce a comprehensive perinatal program:^) family planning and counseling services; (2) genetic screening and counseling services; (3) maternal/fetal transportation; (4) high-risk obstetrical units; and (5) intensive care units for newborns.

Other priorities in the prevention program that promise to have an immediate demonstrable impact on the prevention of mental retardation, or that are cost effective, include regional screening of all newborns for the seven inborn errors previously reviewed and for hypothyroidism and lead poisoning. Provisions must also be in place (via linkages with regional centers) for intervention services such as the PKU diet, further assessment of thyroid status (T-4 testing), and treatment, (for example, BAL treatment for lead poisoning), and so forth. A measles and rubella immunization program, an Rh identification program with subsequent steps included to ensure that

desensitization is provided (availability and administration of RhoGAM), and a vigorous child abuse and neglect prevention program should be implemented.

It should be remembered, however, that the implementation of these priority activities will not entirely prevent the occurrence of mental retardation. With our current incomplete records and knowledge, however, it is important to pursue these activities until all feasible preventive measures are known and/or enacted.

Throughout this presentation, linkages have been suggested between early diagnostic screening services and how or where follow-up treatment can be provided. Priorities must be established so that appropriate intervention can take place. In order to maximally reduce the incidence (that is, the number of new cases in a given year) of the developmental disabilities and have an impact on their prevalence (that is, the number of cases present in a given year), a high priority should be given to the immediate establishment of strong regional combined prenatal and perinatal programs. The goals should be: (1) reduction of the incidence of low birth weight infants; (2) reduction in complications at birth; (3) reduction in complications of the low birth weight infant; and (4) reduction of genetically determined syndromes.

Some of the allied challenges to these goals are: (1) routine rubella titers with follow-up immunization prior to conception; (2) newborn screening for all currently known errors of metabolism via regionalized state supported programs; (3) full funding for regionalized state-wide screening programs for lead poisoning, Rh incompatibility, hypothyroidism, and mercury screening programs; and (4) adequate immunization clinics in each school.

All of these services could be provided by neonatal and perinatal risk centers, located in the major population centers. The rapid availability of genetic counseling (on a regular basis) is an associated necessity. Intervention techniques (alpha-fetoprotein, amniocentesis and, in the future, intrauterine fetal surgery) should also be made available to all who need them.

Further, a high priority must be established for widespread public education for responsible parenthood. To achieve this priority goal, there should be appropriate family planning and counseling available to all persons of childbearing age. The necessary skills and responsibilities of parenthood must be emphasized.

Other services that could be provided by neonatal and perinatal risk centers are:

1. development and support of a state program for family planning under the department of health;

2. increased support of planned parenthood and other women's "walk-in" health clinics to provide contraceptive information and counseling regarding responsible sexual behavior; and

3. acknowledgment that sexuality and family planning education are part of health education and should be actively supplemented by the educational and health care system.

There are a number of other priority areas where a lesser impact can be made in order to decrease the incidence of mental retardation:

1. rapid implementation of an extensive health education curriculum throughout all schools (starting in the first or second grade);

2. programs to prevent child abuse and neglect, including: (a) identification of the family at risk when the mother is pregnant and close follow-up (that is, a maternal childcare nurse who is psychologically sophisticated, can provide a nurturing experience for the parents, serve as a resource person in helping them with childrearing, and foster optimal child development); and (b) use of the nurse (or other developmentally-oriented health care professional) as a liaison with other social-educational-medical service support systems to maximize the services offered to the family—a variety of well-coordinated supportive services over a prolonged period of time do significantly help a large portion of actual or potential abusers;

3. a nutritional education program for all pregnant women; and

4. assessment of the status of catastrophic insurance needs (Major Medical provisions in third-party insurance contracts), so that the outcome of pregnancy is not complicated by unbearable financial burdens to the family.

First Steps

A professional who is committed to advocating prevention-oriented programs in mental retardation should focus on the following "first steps" to effectively aid the citizens in his area:

1. Assure immunization for all infants and preschoolers.

2. Expand program of Rh screening and mandatory marriage license recording of blood typing as soon as possible and include insurance of RhoGAM availability and utilization.

3. Initiate a lead poisoning prevention program by rapidly screening a mass population with the FEP test, treating those currently ill, and spurring the development of rigid lead control measures.

4. Establish at least one regional intensive care unit for at-risk newborns.

5. Establish at least one regional genetic and/or metabolic laboratory to screen more extensively for the inborn errors of metabolism, lead poisoning, and hypothyroidism.

6. Initiate a teenage pregnancy risk center, utilizing currently

available federal funds.

7. Activate and extend local public education programs, especially for primary and secondary school pupils, in the prevention of mental retardation.

8. Extend early intervention education programs (such as Project Homestart and Headstart) to broaden understanding of the psychosocial causes of mental retardation. Support local programs to combat child abuse and neglect.

The current state of the art in prevention of mental retardation has been reviewed. The issues surrounding the establishment of a comprehensive approach to this challenge have been noted. A system for initiating state-wide or regional programs to meet this challenge, including priorities and linkages, has been presented.

Our country has gone a long way toward helping to prevent mental retardation—those disorders that have blighted the lives of millions of its citizens and their families. It can do more. The authors' hope is that this chapter will aid in bringing about a brighter future of full developmental opportunity for all of our country's citizens.

Treatment

Current treatment strategies utilized in the field of mental retardation

can be subsumed under the general subtitles of "Cure for a Few," (for example, early dietary management in phenylketonuria, neurosurgical intervention in craniostenosis), "Treatment For Many" (for example, specific correction of speech and hearing problems, seizure management, psychotherapy for concomitant emotional illnesses), and "Habilitation for All" (for example, special education, vocational training). This section will review the current state of the art by describing possible treatment of the various etiological groupings of the retarded. The following overview of suggested treatments is presented in greater detail by Menolascino and Egger.

Genetic and Chromosomal Disorders

In the chromosomal disorders, cure, as we understand the concept, is not possible because no specific treatment interventions are currently available. At present, for example, strategies of treatment for Down's syndrome consist of: (1) meticulous medical care during the newborn period; (2) close observation for other associated anomalies; (3) early adjunctive treatment for associated disorders (for example, respiratory infections, which frequently require energetic treatment because of anatomical problems in the palate, nose, and sinuses; and (4) a high general vitamin regimen with optional doses of vitamin B and C in particular because of proneness to early and repeated infectious diseases. Down's syndrome patients should also be medically evaluated early in life to establish cardiac status because of the high

prevalence (18 percent) of congenital heart disorders among these patients. During the past ten years advances in techniques of cardiac assessment and surgical intervention have been especially helpful in correcting the cardiac defects in Down's syndrome. These and other treatment approaches can be of great assistance in minimizing the secondary signs of this chromosomal syndrome.

Habilitation strategies can accomplish a great deal in all of the chromosomal disorders (including the cri du chat, trisomy D, and trisomy E syndromes) to maximize the development of these youngsters early in life. These modern habilitation strategies are most clearly observed in recent approaches to Down's syndrome. For example, at the University of Washington, groups of Down's syndrome patients one year of age and older have been taught at an advanced rate a wide variety of perceptual and motor skills (such as tactile and visual-motor differentiation and color discrimination). Progress reports of this eight-year study indicate that most of the Down's syndrome patients in the study are functioning in the mild to borderline range of intellectual ability (rather than the usual severe to moderate intellectional levels seen in nonstimulated Down's syndrome patients)—showing that early habilitation efforts do have a profound impact on the developmental prognosis of patients with major chromosomal disorders. In all the chromosomal disorders, as a general rule, habilitation represents a major resource for amelioration of the effects of these disorders

even though treatment and curative procedures are not currently available.

Disorders associated with sex chromosome abnormalities may or may not also be associated with mental retardation. In both Turner's and Klinefelter's syndromes, endocrine treatment can be very helpful, as can counseling these youngsters and their parents about problems associated with sex differentiation. The disfiguring signs and symptoms of the ears and neck in Turner's syndrome can be corrected by plastic surgery so that these patients appear as normal individuals. Similarly, individuals with Klinefelter's syndrome tend to have a high incidence of cardiac difficulties, and can profit from close attention to factors such as hypertension management and alternation in the level of their physical activity. Although a cure is not available for either Turner's or Klinefelter's syndromes, treatment for physical remediation and supportive psychotherapy may be of considerable benefit by increasing the patient's ability to lead a normal life in the community.

In the congenital neurocutaneous syndromes of childhood (for example, tuberous sclerosis, neurofibromatosis, Sturge-Weber syndrome, etc.) curative strategies of treatment are also not available. However, treatment approaches may be helpful. In tuberous sclerosis, recent work suggests the presence of isolated tubers (firm connective tissue masses) in the brain. Since these tubers seem to be the foci for seizures, their surgical removal has, in selective

instances, dramatically reduced the frequency of seizures. As in all seizure disorders, ongoing neurological consultation and continued care are necessary because the organic basis for the continuing seizure disorder (the tuber) makes these individuals prone to seizure phenomena unless meticulously followed to monitor anticonvulsant status.

Neurofibromatosis patients frequently have disfiguration of the face and neck secondary to the neurofibroma soft tumor masses sited in the myelin sheaths of the cranial nerves of the face and neck. Cosmetic surgery may aid by removing the neurofibromas, and similar surgical treatment can remove the disfiguring facial-skull hemangiomas associated with the Sturge-Weber syndrome. Further, recent treatment advances in the Sturge-Weber syndrome have developed from the observation that calcification of the brain occurs on the same side (hemisphere) as the large papillary hemangioma of the face. In selected instances partial hemispherectomies that remove this calcified brain tissue have significantly decreased the frequency of seizures.

There have been dramatic and significant advances in the cure, active treatment, and habilitation of the mentally retarded associated with the inborn errors of metabolism. Phenylketonuria is the classical example in this area. A child with phenylketonuria whose disease is diagnosed very early and who is placed on a therapeutic diet consisting of synthetic foods specially formulated to be very low in phenylalanine, will tend to develop normally

both physically and intellectually. Recent work suggests strongly that if a mother who has had a previous phenylketonuric child is placed on the PKU diet during her next pregnancy, she will produce a child who has not been excessively exposed to the toxic effects of high phenylalanine levels during gestation. This approach, when coupled with early postnatal dietary treatment, further reduces the risk of damage to the child during the middle years of childhood.

Early diagnosis and treatment, coupled with an appropriate dietary regime, can also ameliorate the effects of maple syrup urine disease. If the diagnosis is made at a sufficiently early age, similar treatment strategies are effective in Wilson's disease. Later in life, treatment of Wilson's disease includes the use of BAL as an aid in precipitating the excess copper out of the system. Similarly, homocystinuria can be effectively treated with dietary supplements of methionine and dosages of vitamins B and C. More active treatment approaches are developing rapidly and, even when such approaches are not curative, they are frequently effective in slowing down the rate of the effects of the inborn error of metabolism.

Both Galactosaemia and fructosuria are examples of defects that can be cured. After early diagnosis, simply switching the child from cow's milk to goat's milk (or a different type of nondairy component) eliminates the deleterious effects of both of these metabolic defects.

The lipid metabolism disorders (for example, Tay-Sachs disease, Spielmeyer-Vogt disease, Niemann-Pick disease) remain an area of concern and active research. Currently there are no available treatment regimens leading to cure. The same can be said in general of the connective tissue disorders such as the Ellis-van Creveld syndrome, Ehlers-Danlos syndrome, and Marfan's syndrome. Here one treats the associated symptoms and uses appropriate supportive habilitation procedures. In the complex inborn errors of sugar metabolism, there is currently a good deal of argument as to whether Hurler's syndrome is curable or not. The enzyme defect in the disorder is known, and some early experimental diets (almost like the early PKU diets) appear to offer the possibility of a cure.

Recent work has shown that youngsters suffering from the Lesch-Nyhan syndrome have a gross abnormality of uric acid metabolism. The high uric acid blood level is associated with major behavioral problems in which they aggressively bite their fingers and lips. Medication similar to that used in treating gout returns the uric acid blood levels to normal and tends to abolish dramatically the behavioral symptoms. Whether these medications will change the long-term course of the disease is not known at this time.

In summary, more and more is being learned about the disorders caused by genetic factors and more and more about specific enzyme defects; this knowledge has provided the medical community with procedures that permit

intervention at a specific biochemical level or dietary level, and similar approaches that actually slow down, if not reverse, the disease process. Many treatment approaches for associated handicaps can appreciably alter the life of the afflicted individuals and greatly assist in ongoing habilitation efforts.

Metabolic Diseases and Intoxications

Metabolic diseases and intoxications leading to the symptom of mental retardation represent, as a general group, disorders that can be controlled and, in selected instances, definitively treated. These disorders represent the result of abnormal levels of enzymes, metabolic substitutes, or derangements of endocrine substances. Primary hypoglycemia, diabetes mellitus, and hypothyroidism are the metabolic diseases most frequently noted during the early years. In the past, without specific treatment, such conditions produced serious medical disease with accompanying brain damage. Now, with therapeutic agents such as insulin, control of the blood sugar level can prevent this disorder from ravaging the brain or other organ systems. Similarly, the early diagnosis and treatment of hypothyroidism (depending on the nature of the enzyme-organ problem involved) is a dramatic medical advance.

Drug toxicity raises the issue of the side effects of particular compounds, such as the risk of deafness associated with streptomycin and the risk of

kidney toxicity secondary to the use of sulfa compounds. The essential strategy in these disorders is to obviate the use of potentially harmful pharmaceuticals when there are other less toxic agents available. For example, returning to the treatment of thyroid disorders, radioactive iodide has been found to be extremely toxic, and perhaps carcinogenic; whereas propylthiouracil can be rather safely utilized. The Thalidomide experience is a clear example of the need for fully understanding the potential for drug Teratogenesis and avoiding such pharmaceuticals in favor of less toxic agents. Preventive postures should also govern the use of the phenothiazines and barbiturates; when such drugs are utilized, the lowest dosage possible should be administered in a fashion that minimally impairs the autonomy of the individual.

There is no specific treatment (and probably never will be) for mental retardation caused by exposure of the fetus to radiation. Prevention is the only effective means of control.

The alkylating agents (such as those for the excretion of lead and mercury) are excellent treatment interventions for individuals exposed to excessive amounts of lead or mercury compounds. The energetic use of BAL factor for decreasing the lead levels in the body can be considered curative for associated symptoms such as the anemia that is produced by this intoxication.

In reducing mental retardation related to maternal diabetes, the optimal treatment approach is meticulous clinical management of the diabetes mellitus in the pregnant mother. It is essential to be on the alert to identify the fragile juvenile diabetic who becomes pregnant and will require close clinical monitoring of her metabolic status.

As a cause of mental retardation, toxemias of pregnancy have been decreasing in frequency as a result of mass screening for hypertension. Treatment is supportive during the acute toxemia state. Toxemias of pregnancy are best approached by preventive measures and meticulous obstetrical care of the mother.

Prenatal, Perinatal, and Postnatal Factors

This is a very complex area since it bridges such diverse disease entities as toxic states, infections, blood incompatibility, and improper diet. Treatment of the multiple factors leading to mental retardation associated with the prenatal, perinatal, and postnatal periods is often a secondary consideration. There are a significant number of general preventive strategies, such as assuring excellent prenatal care, focusing on maternal nutrition, and providing general social and financial support for the mother and her family. These strategies, however, transcend the resources of the usual health care system.

They include social programs that foster the well-being of ail individuals, such as the recent federally funded program to establish centers for pregnant teenagers, not only to give help while they are pregnant, but also to provide counseling and redirection of their life styles, in hopes of preventing future unwanted pregnancies.

Prenatal Factors

Among the specific prenatal causes of mental retardation, are congenital syphilis and rubella. With syphilis, the issue of public education and early diagnosis with specific laboratory testing during the newborn period arises again. If there is an active infection phase in the newborn child, a cure is now possible via penicillin treatment. Treatment of syphilis takes on more specificity in the secondary and tertiary stages of this infection.

Rubella is a stellar example of what can be accomplished in the primary prevention of mental retardation. Recent national public health efforts have produced population immunization levels in the United States of between 92 to 95 percent of females under eighteen years of age. This figure provides great hope that the recurrent epidemics of rubella—and its tragic aftermath for pregnant women—will no longer occur in our country. Yet, even when primary prevention efforts have not been utilized, there is still much that can be done therapeutically after the birth of a child whose mother had rubella

during the first trimester of her pregnancy, and there is ample opportunity for active tertiary prevention programs for the child. The most common residual handicap of congenital rubella, a high frequency hearing loss, can be effectively ameliorated by special programs for the hearing-impaired. Other associated handicaps include mental retardation, microcephaly, and blindness. Programs for the visually impaired can be helpful in dealing with the child's visual problems. These children also have special needs for intensified contact with people, especially early mother-child interactions. Accordingly, a very energetic program to promote personal interaction is needed in the first year of life. Without this psychosocial development intervention, the youngster is likely to develop a fixed autistic orientation to the outside world.

Infectious diseases of both prenatal and postnatal origin continue to present major treatment challenges. For example, though toxoplasmosis continues to be a relatively frequent prenatal syndrome which can produce mental retardation, no specific treatment exists for it at this time. Repeated attempts at a cure have been made with anti-parasitic drugs, but it continues to be extremely resistant to definitive treatment. Antiviral agents have been utilized in another prenatal infection, cytomegalic inclusion body disease, but the findings as to their efficacy are still equivocal. These and similar prenatal disorders continue to present a treatment challenge, whether *in utero* or in the newborn and early infancy periods. Of the childhood diseases, rubella in

particular presents a special challenge. Ironically, many professionals continue to view these disorders as being "usual," "common," or "expected" in childhood. Accompanying this attitude is the opinion that these disorders are not dangerous even though according to clear data, one-third of all children who display clinical signs of rubella tend to have abnormal electroencephalograms during the febrile phase of this disease. They are extremely at risk to develop encephalitis secondary to this systemic viral disorder. Such considerations underscore the need for meticulous medical care during the acute period of infection. Utilization of supportive medical care should be addressed to the use of anticonvulsants and/or antibiotics for the secondary symptoms and infections of the primary generalized rubella infection. This contemporary approach to the active management of rubella can be helpful in preventing secondary manifestations or resultant handicaps.

Perinatal Factors

The perinatal factors that increase the likelihood that a mother will have a retarded child are well known: prolonged labor, toxemia, prematurity, difficult birth, perinatal hypoxia, maternal syphilis, maternal pelvic radiation, maternal diabetes, prolonged maternal infertility, low socioeconomic status, previous birth of a defective child, maternal age (very early adolescence or over age thirty-five years), and high parity (five or more pregnancies, especially if closely spaced). In all of these factors, preventive treatment

should focus on: (1) full awareness and documentation of the fact that the mother is at risk; and (2) maximum prenatal care for the mother. Modern prenatal care represents the best possible means of providing the newborn child as great a chance as possible to develop normally. After birth, the child of the at-risk mother should be fully evaluated and closely observed, with close attention to supportive physical care. For example, physical factors that present major treatment challenges are frequently noted in infants of low birth weight with associated congenital anomalies of the limbs. Special sensory defects of these infants require energetic treatment. From across the world, the findings of the last decade strongly indicate that if early treatment is provided, there is a sharp decrease in both the number and extent of symptoms. For example, infants who have experienced a physically traumatic birth may develop symptoms of cerebral palsy. Treatment techniques, such as developmentally oriented physical therapy, which focuses upon increasing the range of motion of the child's limbs and decreasing spasticity, may well result in the elimination of 75 to 80 percent of cerebral palsy symptoms. These children should be closely monitored by follow-up observation and allied interventions. These treatment strategies also foster early development and habilitation.

Postnatal and Early Childhood Diseases

Postnatal infections continue to be a major cause of mental retardation.

The two major groups of these disorders are the infection processes subsumed under the terms meningitis and encephalitis. It is of crucial importance to remember that these two disorders are entirely different, tend to have distinct infectious agents, and subsequently may require different treatment approaches. Meningitis is an infectious disorder that primarily involves the meninges of the brain, is bacterial in nature, and, if diagnosed early, can be effectively treated by a broad range of antibiotics. Encephalitis, on the other hand, does not specifically involve the meninges. It directly attacks brain substance itself, tends to be caused by viral agents, and is typically not responsive to currently available antibiotic therapy. Effective treatment for meningitis requires: (1) rapid diagnosis; (2) laboratory identification of the bacterial agent involved; and (3) specific treatment interventions which may involve exceeding the usual dosage range of antibiotics. Delayed or ineffective treatment may leave the child with seizures, coma, and mental retardation. Clearly, these meningeal infection disorders are curable. Even those children who are brought in too late for early definitive treatment, usually have far fewer residual handicaps if vigorous treatment and supportive general measures are initiated.

The effective treatment of the infections that cause encephalitis is, at the present time, a most difficult and discouraging problem. There are only a few effective antiviral agents that may be helpful in selected instances of encephalitis. Current treatment emphasizes general physical support during

the period of acute infection to prevent the body from being overwhelmed (for example, cortisone may be needed to prevent adrenal exhaustion), and youngsters who develop seizures during the acute phase need active treatment with anticonvulsants. Active treatment, albeit supportive, increases the rate of cure in encephalitis, but, more importantly, it significantly decreases the number of residual effects of the infectious disease process. Special rehabilitation treatment resources are especially needed for those youngsters who have regressed in their development as a result of a meningitis or encephalitis episode.

Tuberculosis, especially the pulmonary form with brain foci, formerly was endemic in certain parts of the United States. It is rarely seen today. Treatment involves: (1) specific and early diagnosis; (2) specific and early antibiotic therapy; and (3) persistence in the treatment. This treatment regimen has made tuberculosis a minor problem in our country since it is a curable infection. However, if early diagnosis and specific antibiotic agents are not utilized (or not utilized in appropriate amounts), youngsters who have suffered pulmonary tuberculosis may have residual brain damage, special sensory impairments, and mental retardation.

New Growth and Tumors

New growths or neoplasms, which result in the symptom of mental

retardation, encompass disorders that are essentially restricted to the central nervous system. Benign and malignant brain tumors are the most frequent causative agents. Treatment centers around: (1) early diagnosis before the tumor has grown so far that it has destroyed adjacent brain tissue; (2) attempts to remove neurosurgically the abnormal tissue to determine whether or not it is malignant; and (3) support for the child whose world has changed as a result of the tumor, whether it is malignant or not. Accordingly, the major treatment approach is definitive surgery or surgical techniques that focus on aiding the child to recoup as much intact function or ability as possible.

Hypothyroidism

It has become clear, during the last five years in particular, that hypothyroidism in the newborn is probably as frequent as phenylketonuria (that is, it occurs in one in 6 thousand to one in 10 thousand births). Mass screening and laboratory confirmation have made possible accurate checks for hypothyroidism in the newborn nursery and subsequent definitive treatments. Although the spectrum of causology can range from enzyme deficiency to tissue anomalies of the thyroid, it can be said that, if detected early, congenital hypothyroidism is curable; if the diagnosis is not confirmed within the first year of life, it is no longer curable. In this instance, the word "curable" indicates reversal of all the skin, blood, and body changes, and of

mental retardation. It is the latter aspect, development of intelligence, that is nonresponsive to treatment intervention after one year. Even so, one must still inaugurate treatment and management to provide specific metabolic support to the individual (for example, thyroid compounds and/or iodine) so as to salvage as much developmental function as possible.

Today, primary prevention strategies do exist for detecting hypothyroidism. In some states, in which newborn screening for hypothyroidism is mandatory, the disorder is no longer present. Universal application of these primary preventive approaches should cause hypothyroidism to become a disease of the past.

General

Disorders that reflect brain or skull malfunction as a cause of mental retardation encompass brain malformations and specific syndromes of the face and skull. In this area, cure, or primary prevention, is currently not a viable treatment goal for the majority of individuals suffering from these disorders. However, a small number of these disorders are amenable to surgical intervention. For example, in the area of spina bifida with associated disorders such as meningomyelocele, surgery in the first week or two of life allows the physician to open the herniated sac in the posterior sacral area of the spine, carefully resect the tissue around the area of the spinal cord,

replace the spinal cord in the vertebral column, and cover it over with a bone graft. If this particular surgery is not performed early in life, the spinal cord will become adhered to the overlying sacral area tissues and will stay fixed at the point where it is herniated out of the spinal column. Later brain-spinal column growth will literally pull the brain down onto the upper cervical vertebrae and produce serious compression of the brain stem. In a number of these instances, surgical intervention can be a curative procedure. In other instances, resultant bilateral paralysis (distally from the umbilicus) may be secondary to a spinal lesion and, in these cases, a general rehabilitative treatment program similar to those utilized in cerebral palsy is needed.

Craniostenosis usually implies that the sutures of the human skull, which are normally about 80 percent closed by two to three years of age, begin instead to close at nine to ten months of age. At this developmental time (under normal conditions) the growing brain is literally pushing the overlying, and still very mobile, skull to ever greater expansion. If the sutures close prematurely, the rapidly enlarging brain begins to fill a closed cranial vault with resulting secondary pressure and atrophy of the underlying brain. The treatment of choice is early diagnosis by means of radiographic studies of the skull. Neurosurgical intervention involves crushing the suture lines, placing strips of platinum or polyethylene in the suture lines, and allowing this material to remain in place until the child is almost three years of age. For many children with craniostenosis this neurosurgical procedure is curative.

Over the past thirty years, great strides have been made in the treatment of hydrocephalus. This condition is caused by production of an excess amount of cerebrospinal fluid in the midline structures of the brain. This excess fluid balloons out the inside of the brain and pushes the skull out to form a very large external configuration. Following radiographic studies to locate and categorize the nature and/or cause of the individual case, a Holtner valve can neurosurgically be placed in one of the lateral ventricles of the hydrocephalic brain and connected to surgical tubing that exits out the adjacent mastoid or is threaded under the skin to the heart or one of the kidneys. The Holtner valve is arranged so that when a certain pressure gradient of the cerebrospinal fluid is reached inside the skull, the valve opens and discharges excess fluid into the mastoid, heart, or kidney for excretion. The use of the Holtner valve and associated surgery can be regarded, in many instances, as curative for hydrocephalus.

Other disorders, such as the first arch syndrome, Rubinstein-Taybi syndrome, and the Klippel-Feil syndrome, are not curable or effectively treatable at this time. However, as with many other disorders, treatment can provide general physical support. Habilitation efforts for the child should be started very early in life.

Psychiatric Disorders

Emotional disturbances in the retarded continue to be a frequent cause for their institutionalization. A study of emotional disturbance in a sample of institutionalized individuals with Down's syndrome noted that whereas only 37 percent of the total sample were emotionally disturbed at the time of the study, 56 percent had displayed significant symptoms of emotional disturbance at the time of their initial admission to the institution. One might ask why most of these people who were emotionally upset were sent to an institution for the retarded rather than to a mental health facility. In contrast, a brief period of inpatient or outpatient psychiatric care, coupled with revised expectations of the parent and increased support in the community, could frequently have prevented institutionalization.

Compared with the incidence of emotional illness in the general population, the retarded are more at risk. Early prevalence studies (before i960) were carried out primarily in institutional or hospital settings, and frequency rates reported ranged from 16 to 40 percent.- A recent series of reports on mentally retarded persons living with their primary families or in community-based settings at the time of study have appeared during the last fifteen to twenty years. These studies, especially those focusing on retarded children under the age of twelve years, have rather consistently reported a 20 to 35 percent frequency of emotional disturbances.-' These findings are especially important when one notes that epidemiological studies on mental illness, for example, the Mid-Town Manhattan and Sterling County studies, in

the general population reveal an incidence of 14 to 20 percent.

In the broadest sense, one finds that the mentally retarded develop essentially the same types of emotional illness that befall persons of normal intellectual ability. One finds in the retarded the full range of psychoses, neuroses, personality disorders, behavior disorders, psychophysiologic disorders, and transient situational disturbances that are noted in the "normal" population.

In community-based psychiatric programs that treat the retarded (community mental health centers), it is not unusual to note combined diagnoses, such as childhood schizophrenia and moderate mental retardation, or unsocialized aggressive reaction of adolescence and mild mental retardation. Practically speaking, certain diagnostic categories, such as the neuroses, tend to be underrepresented in the retarded, while others are seen with relative frequency (for example, schizophrenia, the various behavioral reactions, and transient situational disturbances). The following section reviews the diagnostic entities seen most frequently and those that present special problems of diagnosis and treatment.

Psychotic reactions of childhood have presented a major challenge to the clinicians since they were recognized as distinct in the first decade of this century. Delineation of types and etiologies has been delayed, in part, by the

fact that the psychotic child frequently functions at a mentally retarded level, and early observers believed that all psychotic children deteriorated. In 1943, early infantile autism was described and became the focus of much interest, including speculation as to whether it represented the earliest form of childhood schizophrenia. The term "autism" is frequently employed in the differential diagnosis of severe emotional disturbances in infancy and early childhood. Yet, to label a child autistic presents some formidable problems with regard to definition of the term, specific etiological-diagnostic implications, and treatment considerations. All too often, the word is used as if it were a diagnosis, a synonym for childhood schizophrenia, or an abbreviation for early infantile autism. Such usage obviously is imprecise and contributes further to the diagnostic confusion that has abounded in the literature concerning childhood psychosis.

Interestingly, today there is not the degree of fervor over diagnosis, treatment, and differential outcome concerning the functional childhood psychoses and their interrelationships to mental retardation that existed ten to fifteen years ago. A number of follow-up studies, the rediscovery of the wide variety of primitive behavioral repertoires in the retarded (the same behavior that had been termed "psychotic" in the past), and a lack of relative differences in treatment modalities and corresponding responses to them, have all tended to mute the intensity of this earlier clinical debate. For example, an excellent review of the past relationships between emotional

disturbance and mental retardation by Garfield and Shakespeare addressed almost a third of its content to the relationships between emotional disturbance and mental retardation. Indeed, as Creak and Penrose have noted, the most common challenge is to ascertain not whether the patient is retarded or psychotic, but how much of his condition is attributable to retardation and how much to psychosis.

Earlier it was noted that the psychoses of childhood intensified the study of the interrelationships between mental retardation and the psychoses of childhood. Now in the 1980s, the issue has been clarified, and it is becoming apparent that the number of functional etiologies of infantile autism and childhood schizophrenia is limited. The reported evidence for central nervous system pathology in the psychoses of childhood is the most frequent trend noted in the past ten years.

In summary, these clinical reports have shown clearly that: (1) the psychoses of childhood, particularly autism, are strongly associated with dysfunction of the central nervous system; (2) the appearance of psychotic behavior (and/or autistic behavior) and mental retardation in young, nonverbal children indicates both common etiology and a diminished capacity to tolerate stress; (3) retarded patients may show stereotyped, self-stimulating behavior that resembles autism; and (4) relief of the psychotic condition in “autistic” children far more commonly produces a retarded child

who is able to interact with others rather than a child of normal intelligences.

Personality disorders are characterized by chronically maladaptive patterns of behavior (for example, antisocial personality, passive-aggressive personality, and so forth) that are qualitatively different from psychotic or neurotic disorders. Studies reported in the earlier history of retardation tended indiscriminately to see antisocial behavior as an expected behavioral accompaniment of mental retardation. Indeed, the much discussed earlier reports on the relationship between retardation and personality disorders — especially the antisocial personality—were couched in moralistic-legal terms rather than containing definitive descriptive criteria. The antisocial personality designation continues to receive much attention, and it is frequently overrepresented in references to borderline and mildly retarded individuals. It would appear that, for a variety of reasons, behavioral problems of an antisocial nature are more frequently seen in this group. The same poverty of interpersonal relationships during childhood that leads to retardation associated with psychosocial deprivation can also lead to impaired object relations and poorly internalized behavioral and emotional controls. Also, the diminished coping skills of this group often lead them to perform deviant acts simply to exist. Finally, this group is most likely to be released from institutional settings in young adulthood, and their behavior illustrates graphically the effects of institutionalization on personality structure. It is interesting to note that other personality disorders (for

example, schizoid personality) have been reported only rarely in the retarded. Indeed, the only other personality disorder in the retarded that has received much attention is the "inadequate personality," even though the application of exact diagnostic criteria would exclude this disorder as a primary diagnosis in mental retardation.

In summary, although personality disorders do occur in the mentally retarded, they are based primarily on extrinsic factors, have no distinct etiological relationships to mental retardation, and, despite persistent folklore, are not abnormally frequent in the noninstitutionalized retarded population.

Psychoneurotic disorders were rarely included in discussion of the frequency and types of emotional disturbances in the mentally retarded before 1950.

In 1970, a study disputed the concept of incompatibility between neurosis and retardation. Neurotic phenomena tend to be associated with atypical developmental patterns in conjunction with disturbed family functioning. Psychoneurotic disorders of retarded children tend to be linked to symptoms of anxiety, fear of failure, insecurity, and to exogenous factors such as chronic frustration, unrealistic family expectations, and deprivations. Psychoneurotic disorders are more common in children functioning in the

high-moderate and mild ranges of mental retardation. This finding has prompted speculation as to whether the relative complexity of psychoneurotic transactions is beyond the adaptive limits of the severely retarded.

Transient situational disturbances are a rather large category of minor emotional disturbances that are perhaps underutilized in the clinical assessment of emotional disturbances in the retarded population. The authors think that this underutilization is one of the major drawbacks of descriptive approaches to the retarded. It is defined as a category reserved for transient disorders of any severity (including those of psychotic proportions) that occur in individuals who have no apparent underlying mental disorders and that represent an acute reaction to overwhelming environmental stress. The transient nature of these disorders is their paramount feature, and the assessment of adaptive capacity poses a recurrent dilemma when one works with a retarded population. If the clinician thinks that retarded people have poor adaptive capacities and, therefore, expects little resolution, treatment intervention is less than energetic, and other diagnostic categories are often utilized. Furthermore, if the mentally retarded are considered to be excessively prone to emotional disturbances, "transient" is then viewed as the beginning of a chronic emotional disturbance that has emerged to accompany the retardation.

In the authors' experience, a great number of emotional and behavioral problems in the retarded are transient in that they are frequently caused by inappropriate expectations or rapid changes in life patterns. These problems often respond rapidly to environmental adjustments. In summary, even though it is not possible to use the transient situational diagnosis if one follows the letter of current diagnostic guidelines, it is hoped that professionals will conceptualize disorders in the retarded in this manner when it is appropriate.

Problems Associated with Different Levels of Retardation

The severely retarded are characterized by gross central nervous system impairment, multiple physical signs and symptoms, and a high frequency of multiple handicaps (in particular, special sensory and seizure disorders). Such severe problems directly impair their ability to assess and effectively participate in ongoing interpersonal-social transactions. Clinically, these patients manifest primitive behaviors and gross delays in their developmental repertoires. Studies by Chess, Korn, and Fernandez of severely retarded children with the rubella syndrome and by Grunwald on the multiply handicapped-severely retarded clearly document the high vulnerability of these children to psychiatric disorders.

From a diagnostic viewpoint, the very primitiveness of the severely

retarded person's overall behavior, in conjunction with much stereotyping and negativism, may be misleading. For example, when under minimal stress in an interpersonal setting, mentally retarded children frequently exhibit negativism and out-of-contact behavior. This behavioral response may initially suggest a psychotic disorder of childhood. However, these children do make eye contact and will interact with the examiner quite readily, despite their minimal behavioral repertoire.

The authors have been impressed by the extent of personality development the severely retarded can attain if early and energetic behavioral, educational, and family counseling interventions are initiated and maintained. True, they remain severely handicapped in their cognitive and social-adaptive characteristics, but there is a world of difference between the severely retarded child with many self-help skills who graduates from a standing table to a wheelchair and the untrained, severely retarded one who tries to withdraw from, or is aloof to, interpersonal contacts and who is totally lacking in self-help skills. Interestingly, these youngsters tend to be accepted by their parental support systems and peer groups (if adequate evaluations and anticipatory counseling are accomplished), perhaps reflecting empathy for the obvious handicaps they display.

The moderately retarded encompass some of the same etiological dimensions noted previously accompanied by a wide variety and high

frequency of associated handicaps. The children's slow rate of development and their specific problems with language elaboration and concrete approaches to problem-solving situations present both unique and marked vulnerabilities. In an outstanding study, Webster viewed these personality vulnerabilities as stemming from the characteristic postures moderately retarded children tend to use in their interpersonal transactions: more autism (selective isolation), inflexibility, repetitiousness, passivity, and a simplicity of emotional life. This simplicity of emotional life, a cardinal characteristic of the moderately retarded, reflects their undifferentiated ego structures and poses the clinical challenge of attempting to modulate their tendency toward direct expression of basic feelings and wants, as noted in their obstinacy, difficulties in parallel play situations, and so forth. The limited repertoire of personality defenses, coupled with their concrete approaches, tends to be fertile ground for overreaction to minimal stresses in the external world. Proneness to hyperactivity and impulsivity, rapid mood swings and temporary regression to primitive self-stimulatory activities are characteristic of their fragile personality structures. Unlike the severely retarded, these youngsters tend to be rejected by their parents and peers. Their significant attempts to approximate developmental expectations, coupled with the aforementioned behavioral traits, appear to alienate them from those very interpersonal contacts they so desperately need.

The mildly retarded have given rise to debate as to whether they

represent the statistical expression of the polygenetic basis of the symptom of mental retardation, or whether they are the untutored "have-nots" of a society that tolerates only minor deviations from the norm. Emotional disturbances of the mildly retarded reflect the well-known residuals of a person who is labeled deviant and then becomes caught in the dynamic interplay of disturbed family transactions. The typical delay in establishing that these youngsters have a distinct learning disability (usually not confirmed until six to nine years of age) presents the mildly retarded individual with a constant source of anxiety about his inability to integrate the normal developmental sequences at the appropriate time in his life. Usually, during the latent period of psychosexual personality integration, mildly retarded children have considerable difficulty in understanding the symbolic abstractions of schoolwork and the complexities of social-adaptive expectations from both family and peer group. Often at this stage they gain some insight into their limitations and, by early adolescence, have established an identity that incorporates both retardation and deviance. Frequently the vulnerabilities of the mildly retarded are not buffered or redirected by loved ones into new interpersonal coping styles to help correct earlier misconceptions about the self. Without some community support and direction, the mildly retarded are at high risk for failure in society —especially urban society. In the past, if they managed to avoid an institutional setting for the retarded, it was not unusual to find them, eventually, in other types of institutions, such as correctional

facilities or state-supported psychiatric hospitals.

In summary, it appears that mildly mentally retarded individuals are very likely *not* to be readily identified as handicapped and needing support. Rather, they are seen as society's misfits who, if not simply ridiculed, are apt to be taken advantage of in far more serious ways because of their lack of judgment and limited coping skills.

Psychiatric Problems Associated with Different Models of Care

Another way to conceptualize the problems of the retarded, in addition to types of emotional disturbance and levels of retardation, is by problems that appear to be related to different models of care. Providing treatment guidelines for optional care for the retarded at home, in the community, or in an institutional setting is extremely difficult since there is no such thing as an "average" retarded child. In a general way, they can be grouped by overall abilities; but one of the most striking things about the retarded is the great variation in abilities that may be seen in each individual. This variability, plus the great difficulty caregivers are likely to experience in attempting to understand fully the retarded person's abilities and disabilities, appears to be the basis of a number of the psychiatric problems seen in the retarded. The most common type of error in the care of the retarded is a result of the caregiver's expecting either too little or too much. Too few expectations,

combined with too little effort on the part of the care providers, is often the lot of formerly institutionalized retarded individuals. They tend to show a pattern of underachievement and a detachment syndrome that is typical of people reared in barren institutional environments. One common characteristic is a profound and often indiscriminately expressed affect hunger. Because they often have not had experience with significant or meaningful object relations and have been accustomed to living amid large numbers of minimally involved people, their indiscriminate approach to strangers may lead to serious problems. Another variant is the situation in which the caretaker actually does too much instead of too little—an overprotective model. Parents sometimes feel that the only acceptable solution is to keep their retarded child or adult at home, and too often they are assigned to an isolated part of the home away from the bulk of family or external social contacts. Here, the family caters to their retarded relative's every need and, in doing so, increases his dependence and almost totally eliminates his ability to develop effective social-adaptive functions.

In summary, while the detached, mildly or moderately retarded person is at risk of becoming the counterpart of a person of normal intelligence with a character disorder, the overprotected retarded person is likely to show symptoms of inflexibility, autistic thinking, and situational anxiety.

At the other end of the spectrum, there are retarded individuals who

show clear evidence of their caretakers' having expected too much from them. One of the most common problems in very young, moderately retarded children who do not have physical stigmata is a failure by the parents to recognize their children's intellectual limitations before the normal time for language acquisition. It would appear that one common cause of autistic-like psychoses is the placement of a sensitive, intelligent-appearing, but nevertheless retarded, child in a situation in which conscientious parents are doing all the "right" things during the second year of life to facilitate language skills. Verbal demands often cause the moderately retarded child with a language disability to react with increasing anxiety and a variety of avoidance behaviors that result from his lack of pleasure in verbal interactions. Similar examples of excessive expectations are occasionally seen in innovative institutional or community programs in which children who are severely retarded may be urged into overintense efforts to maximize their capabilities. In some cases this results in more frequent seizures, and in others produces a pattern of autistic withdrawal similar to that noted previously. One of the most distressing problems with older children in this group is outbursts of violent behavior when excessive expectations have been maintained too long. All too often such children are placed on high doses of medication in an effort to control aggression that is actually reactive in nature and not a symptom of major emotional disturbance.

Comprehensive Approach to Treatment

A comprehensive-treatment approach to mentally retarded children with associated emotional disturbances should include the following basic principles.

Open-minded Approach

The diagnosis and treatment of children who are both mentally retarded and emotionally disturbed necessitate an open-minded approach. This is the first basic requirement for the clinician who plans treatment for these youngsters, and it is important to maintain this approach throughout treatment. Periodic reevaluation often reveals developmental surprises that underscore the need for a flexible diagnostic-prognostic attitude.

Active Family Participation

The second principle in planning treatment for children with both mental retardation and emotional disturbance is to engage the family in active participation as early as possible. The family is the key to any effective treatment program. The clinician's attitudes and level of interest frequently determine the success of this endeavor; thus, future cooperation with the family (or lack of it) may reveal his unspoken, as well as spoken, attitudes at the time of initial contact. The therapist should convey to the family his willingness to share the facts of the case—not at the end of treatment, but as

part of the first step. Treatment plans must be a cooperative process that parents and clinician work out during the course of treatment.

It is valuable to indicate at an early contact that treatment planning rarely results in a single recommendation; it may shift in focus and alter its course as the child grows and develops. Diagnostic and treatment flexibility in the early stages help a clinician to view the total child and refer to other special sources of help as required.

Much has been written about the grief reactions of families with handicapped children. Such a reaction frequently occurs in parents of mentally retarded children. The clinician evaluating these children must be alert to this grief reaction when first offering parents an interpretation of their child's condition and in subsequent interviews.

Assessment of family interactions and strengths is a necessary part of the total evaluation, since these assets are essential to planning a comprehensive treatment program. Conversely, some of the family psychopathology may reactivate the difficulties of the child being treated. Several interviews may be necessary to determine the nature of family transactions.

Early Diagnosis and Treatment

A third principle of the comprehensive treatment approach is early descriptive diagnosis and early treatment. This includes clarification not only of what needs treatment but also of what can and cannot be actively treated. Full discussion of therapeutic goals will assist families in establishing realistic treatment expectations so that mutual frustration is reduced and fewer secondary psychiatric problems are encountered. In this sense, prevention becomes part of the ongoing work with the child and his family. This total approach requires continued follow-up of the patient. Periodic reevaluations must be done, and appropriate shifts in treatment and expectations carried out.

Initial Contact

The fourth principle is to accept each child as he is at the time of initial contact. He needs acceptance for what he is, not what he might have been without his problem or if therapy had been undertaken sooner. A corollary of this principle is awareness of the family's feelings and acceptance of them as they are. Increasing the parents' guilt feelings is rarely, if ever, desirable in attempting to motivate them toward therapy.

Maximization of Developmental Potential

The fifth principle requires focusing on the maximization of developmental potential. It calls for a different type of goal setting from the

usual treatment expectation, since the focus often must be on what the child can do rather than on an anticipated cure. The goal then becomes one of trying to provide the child with the necessary opportunity and support to develop as fully as possible and to reduce obstacles to a minimum.

Coordination of Services

The sixth principle is to coordinate the services needed by the child. This requires awareness of the various services available in a given community and an attitude that permits collaboration. It necessitates sharing the overall treatment plan with the child (when his or her overall coping ability permits) and the parents. As with other groups of dependent psychiatric patients, child or geriatric, the clinician's efforts must frequently be directed as much toward assessing the strengths and weaknesses of the family and coordinating the available community support systems, as they are toward care of the individual patient. Many retarded people with acute emotional problems are institutionalized unnecessarily. In nearly all cases, the retarded can be maintained in the community with the help of mental health personnel who are willing to provide short-term and long-term care for them.

Bibliography

Acosta, P. "Prenatal Supplements and Breast Feeding," in Koch, J., ed., *International Summit on*

Prevention of Mental Retardation from Biomedical Causes, President's Committee on Mental Retardation, Washington, D.C.: Department of Health, Education and Welfare, Pub. No. HDS 78-21023, 1978, pp. 111-135.

American Academy of Pediatrics. *Report of the Committee on Infectious Diseases,* 18th ed. Evanston, Ill.: American Academy of Pediatrics, 1977.

Barr, M. W. *Mental Defectives, Their History, Treatment and Training.* Philadelphia: P. Blakiston, 1904.

Beattie, A. D., et al. "Role of Chronic Low Level Lead Exposure in the Etiology of Mental Retardation," *Lancet,* 1: 7907 (1975): 589-592.

Beeson, P. B., and McDermott, W., eds. *Textbook of Medicine,* 14th ed. Philadelphia: W. B. Saunders, 1975.

Berry, H. K., et al. "New Approaches to Treatment of Phenylketonuria," in Mittler, P., ed., *Research to Practice in Mental Retardation,* vol. 3. Baltimore: University Park Press, 1977, pp. 229-239.

Birch, H. G., and Gussow, J. D. *Disadvantaged Children: Health, Nutrition, and School Failure.* New York: Grune & Stratton, 1970.

California Association for the Retarded. *Prevention: An Agenda for Action.* Sacramento: California Association for the Retarded, 1977.

Chess, S. "Emotional Problems in Mentally Retarded Children," in Menolascino, F. M. ed., *Psychiatric Approaches to Mental Retardation.* New York: Basic Books, 1970, pp. 55-67.

Chess, S., Korn, S., and Fernandez, P. B. *Psychiatric Disorders of Children with Congenital Rubella*. New York: Brunner/ Mazel, 1971.

Collins, D. T. "Head-banging: Its Meaning and Management in the Severely Retarded Adult," in Menolascino, F. J., ed., *Psychiatric Aspects of the Diagnosis and Treatment of Mental Retardation.* Seattle: Special Child Publications, 1970.

Cooke, R. Personal communication, November 16, 1978.

Creak, E. M. "Childhood Psychosis: A Review of 100 Cases," *British Journal of Psychiatry,* 109 (1963): 84-89.

DeSanctis, S. "Sopra Alcune Varieta Della Demenza Precoce," *Rivista Sperimentale di Freniatria e di Medicina Legale in Relazione con l'Antropologia e le Scienze Gueridiche et Sociale*, 32 (1906), 141-165.

Eisenberg, L. "Caste, Class, and Intelligence," in Murray, R. F., and Rossner, P. L., eds., *The Genetic, Metabolic, and Developmental Aspects of Mental Retardation.* Springfield, Ill.: Charles C. Thomas, 1972. pp. 185-198.

Feingold, M. "Amniocentesis: For Whom, By Whom?" *Patient Care,* 11:20 (1977): 16-61.

Fotheringham, J. B., and Morrison, M. *Prevention of Mental Retardation.* Toronto: National Institute of Mental Retardation, 1976.

Garber, H., and Heber, F. R. "The Milwaukee Project—Indications of the Effectiveness of Early Intervention in Preventing Mental Retardation," in Mittler, P., ed., *Research to Practice in Mental Retardation,* vol. 1. Baltimore, Md.: University Park Press, 1977.

Garfield, A., and Shakespeare, R. "A Psychological and Developmental Study of Mentally Retarded Children with Cerebral Palsy," *Developmental Medicine and Child Neurology,* 6 (1964): 485-89.

General Accounting Office. *GAO Plans Federal Effort to Prevent Retardation.* Washington, D.C.: General Accounting Office, 1977.

Ghadimi, H. "Aminoacidopathies and Mental Retardation," in Murry, R. F., and Rossner, P. L., eds., *The Genetic, Metabolic, and Developmental Aspects of Mental Retardation.* Springfield, Ill.: Charles C Thomas, 1972, pp. 45-57.

GRUNWALD, K. "International Trends in the Care of the Severely and Profoundly Retarded and Multiply Handicapped," in Menolascino, F. J., and Pearson, P. H. eds., *Beyond the Limits: Innovations in Services for the Severely and Profoundly Retarded.* Seattle:

Special Child Publications, 1974.

Guthrie, R. "A Position Paper: Undernutrition in the Infant as a Cause of Mental Retardation," *Mental Retardation News,* 19:10 (1970): 2.

----. "Current and Newer Aspects of Newborn Screening," in Mittler, P., ed., *Research to Practice in Mental Retardation,* vol. 3. Baltimore: University Park Press, 1977, pp. 13-19.

Hayden, A. H., and Hering, N. G. "The Acceleration and Maintenance of Developmental Gains in Down's Syndrome School-age Children," in Mittler, P., ed., *Research to Prevention in Mental Retardation,* vol. 1. Baltimore: University Park Press, 1977.

Hollingshead, A. B., and Redlich, F. C. *Social Class and Mental Illness*. New York: John Wiley, 1958.

Huseman, C. A. "Neonatal Thyroid in Screening for the State of Nebraska," *Nebraska Medical Journal,* 63:8 (1978): 278-280.

Jervis, G. "The Mental Deficiencies," in Arieti, S. ed., *The American Handbook of Psychiatry,* vol. 1. New York: Basic Books, 1959. pp. 1289-1316.

Jones, K. L., Smith, D. W., and Hanson, J. W. "The Fetal Alcohol Syndrome: Clinical Delineation," *Annals of the New York Academy of Science,* 273 (1976): 130-139.

Kanner, L. "Autistic Disturbances of Affective Contact," *Journal of the Nervous Child.,* 2 (1943): 217-50.

----. "Feeblemindedness: Absolute, Relative and Apparent," *Journal of the Nervous Child,* 7 (1948): 363-97.

Koch, J., ed. International Summit on Prevention of Mental Retardation from Biomedical Causes, President's Committee on Mental Retardation, Washington, D.C.: Department of Health, Education and Welfare, Pub. No. HDS 78-21023, 1978.

McCoy, K., "Report on the Wingspread Conference on Adolescent Sexuality and Health Care," *Journal of Clinical Child Psychology,* 3:3 (1974): 18-22.

MacCready, R. A. "Rh Hemolytic Disease Prevention in the United States," Paper presented at Meeting of the American Association on Mental Deficiency, Chicago, Ill., May 3, 1977.

Maravilla, A. "The Early and Periodic Screening, Diagnosis, and Training Program," *Clinical Pediatrics,* 16 (1975): 173-178.

Menolascino, F. J. *Challenges in Mental Retardation: Progressive Ideology and Services*. New York: Human Sciences Press, 1977.

----, and Bernstein, N. R. "Psychiatric Assessment of the Mentally Retarded Child," in Bernstein, N. R., ed., *Diminished People.* Boston: Little, Brown, 1970, pp. 201-222.

Menolascino, F. J., and Eaton, L. "Psychoses of Childhood: A Five-year Follow-up Study of Experiences in a Mental Retardation Clinic," *American Journal of Mental Deficiency,* 72 (1967): 370-380.

Menolascino, F. J., and Egger, M. J. *Medical Dimensions of Mental Retardation.* Lincoln, Neb.: University of Nebraska Press, 1978.

Michigan Department of Public Health. *Hypothyroid Mass Screening.* Lansing, Mich.: Michigan Department of Public Health, 1978.

Milunsky, A. *Know Your Genes*. Boston: Houghton, Mifflin, 1977.

Mittler, P., ed. Research to Practice in *Mental Retardation,* vol. 3. Baltimore: University Park Press, 1977.

National Academy of Sciences. *Maternal Nutrition and the Course of Pregnancy.* Washington, D.C.: National Academy of Sciences, 1970.

Ornitz, E. M., and Ritvo, E. R. "The Syndrome of Autism: A Critical Review," *American Journal of Psychiatry,* 133 (1976): 609-621.

Penrose, L. S. "The Contribution of Mental Deficiency Research to Psychiatry," *British Journal of Psychiatry,* 112 (1966): 747-755.

Perino, J., and Ernhart, C. "The Relation of Sub-clinical Lead Level to Cognitive and Sensorimotor Impairment in Black Preschoolers," *Journal of Learning Disabilities,* 7:10 (1974): 616-620.

Phillips, I., and Williams, N. "Psychopathology: A Study of 100 Children," *American Journal of Psychiatry,* 132 (1975): 1265-1273.

Piomelli, S. "A Model Program for Lead Screening," in *International Summit on Prevention of Mental Retardation from Biomedical Causes.* Washington, D.C.: Department of Health, Education and Welfare, 1979.

President's Committee on Mental Retardation. *Islands of Excellence,* Washington, D.C.: U.S. Government Printing Office, 1973.

Rosso, P. "Nutrition, Brain Growth, and Prevention of Mental Retardation," in Koch, J., ed., *International Summit on Prevention of Mental Retardation from Biomedical Causes, President's Committee on Mental Retardation,* Washington, D.C.: Department of Health, Education and Welfare, Pub. No. HDS 78-21023,1978, pp. 98-110.

Rutter, M., and Schopler, E., eds. *Autism: A Re-appraisal of Concepts and Treatment.* New York: Plenum Press, 1978.

Vermont Department of Public Health. Mental Retardation Prevention Plan. Burlington, Vt.: Vermont Department of Public Health, 1977.

Webster, T. G. "Unique Aspects of Emotional Development in Mentally Retarded Children," in Menolascino, F. J., ed. *Psychiatric Approaches to Mental Retardation.* New York: Basic Books, 1970, pp. 3-54.

Wynn, M., and Wynn, A. *Prevention of Handicaps of Perinatal Origin.* London: Foundation for Education and Research in Child Bearing, 1976.