Developmental Retardation and the Family Physician

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The formidable problem of the family with a developmentally retarded child is discussed with particular reference to the role of the family physician as organizer-coordinator of the highly complex multidisciplinary requirements of this medical problem. A case is presented illustrating numerous aspects of diagnosis and management, as well as pitfalls to be avoided. The quality of the family physician as one who is concerned, accepting, open and honest, as well as accessible to the families under his care, is taxed to the fullest in this instance. Regular joint family visits, together with monitoring of this problem during routine medical visits, are effective means of anticipating and meeting the family's needs.

The family physician in everyday practice faces three major kinds of problems: 1) health problems which he can definitively manage; 2) problems of varying frequency and such complexity that the help of consultants and community resources is required; and, 3) uncommon or rare problems which involve the family physician primarily in early diagnosis, emergency care and family adaptations. The family including an individual with developmental retardation represents the second of these categories. There are few conditions in clinical medicine which encompass problems of such broad range or require such a multidisciplinary approach as the child with developmental retardation.

Mental retardation is said to involve approximately three percent of our population.[1] It therefore represents a significant clinical problem worthy of the family physician's attention in some depth. This paper will examine the role of the family physician caring for a patient and family with this problem, present an illustrative case report, and discuss the opportunities as well as pitfalls of interventions by the family physician.

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Should the Family Physician Become Involved?

The physician frequently becomes apprehensive for a number of reasons when confronted with a child with either mental or physical handicaps such as mental retardation or cerebral palsy. He may consider the prognosis poor and therefore reject the problem as hopeless. He may see the problem as a long-term, highly time-consuming endeavor with less tangible rewards than the solution of an acute clinical process. More important, he may feel ill-equipped to manage such a problem because of his lack of familiarity with the specific, highly-sophisticated skills which must be employed in the proper management of such a family.[2]

The family physician should be willing to assume the dominant role in the long-term evaluation and management of such a child for these reasons:[3]

1. He can contribute the unique perspective of the fam-

ily physician in assuming the long-term continuous care of those families who are so afflicted.

2. He identifies the family as his patient and not simply the afflicted member. He is, therefore, constantly aware of the impact of the clinical problem upon the entire family.

3. He has already assumed the role of advocate in the medical care system for this family. The long-term management of the child with developmental retardation requires, more perhaps than most other clinical problems, such an advocate.

4. As the long-term point of entry into the medical care system for the family, he will be the one continuous thread over many years to coordinate the various modalities of care necessary. He will be seeing the particular patient for routine visits, and in so doing will maintain complete medical records, including all consultation reports.

The family physician must be knowledgeable about the high risk elements of those pregnancies he attends, monitoring such variables as a family history of neurologic disease, potential chromosomal aberrations (such as Down's syndrome in the 40-year-old female), family history of inborn errors of metabolism, and social-cultural factors which are frequently associated with a high incidence of mild mental retardation. He must be aware of teratogenic conditions such as rubella in the first trimester, exposure to radiation, blood group incompatibilities, and the presence of many other factors which can give rise to brain damage. Similarly, he must be alert to perinatal and postnatal factors which are potentially harmful to the newborn infant. Events involving potential brain damage such as trauma, intoxication, severe infection and malnutrition should be recognized for their possible significance.[4]

In assuming care of the newborn, a system of developmental appraisal, such as the Denver Developmental Screening Test, should be employed to allow early diagnosis of deficiencies.[5] Through awareness of the common incidence of mental retardation and related risk factors, as well as alertness to variations from normal developmental patterns, the family physician of necessity assumes the clear responsibility in the recognition of the developmentally retarded. His role only begins here, however, since the uniqueness of his relationship with those families who come to him makes him the logical one to assume long-term responsibility for medical management.

Case Report

On May 15, 1970, a 5 lb., 2 oz. male infant (E.S.) was born to a 28-year-old primigravida mother after 42 weeks gestation. The pregnancy was uncomplicated except for mild third trimester edema. Labor was induced and delivery accomplished after approximately 11 hours with vertex presentation. Respiration was delayed approximately seven minutes following delivery; Apgar rating was five at an unknown time after birth. Resuscitative measures were successful and the baby did well until 18 hours of age when he developed repeated episodes of apnea and cyanosis. Blood glucose was 44 mg percent and calcium 8.3 mg percent at this time. The baby was transferred to Le Bonheur Children Hospital, Memphis, Tennessee, under the Department of Pediatric Neurology, University of Tennessee. He was quite depressed and lethargic, without spontaneous activity. The cry was normal but grasp and sucking reflexes were poor and the Moro reflex was absent. Brisk deep tendon reflexes without clonus were present, together with generalized hypotonia. Skull and chest films were normal. Electroencephalogram was abnormal while recording two clinical seizures; a multiplicity of abnormalities were present, suggesting a diffuse cerebral process. Titers for rubella, herpes simplex, CID and toxoplasmosis were negative, as were blood cultures. Lumbar puncture revealed a few white cells, normal protein and sugar. The child was treated with Penicillin, Kanamycin, Dilantin and pyridoxine. Apneic spells continued for two days; normal activity and tone did not occur for seven days. There was some increased spread of sutures by palpation. Bilateral subdural taps were performed but were negative. This boy was discharged with the diagnosis of convulsive disorder, etiology unknown, and was put on chronic Dilantin therapy. There was no family history for neurologic disease or convulsive disorder for either the father or the mother.

At six months of age, E.S. was evaluated by the Department of Pediatric Neurology at the University of California San Francisco where it was noted that he had the overall appearance of a two to three month old infant. Physical examination revealed a mild strabismus on the left and a generalized increase in muscle tone, greater in the legs than in the arms, but with heel cord tightening bilaterally. There was no evidence of disorder of movement. Sensory examination was normal. Head circumference was small at 15¼ inches (below the tenth percentile for age). Developmental retardation was present without recurrence of convulsions. A very poor prognosis was given to the parents which caused them great distress and considerable resentment. The boy was returned to a local pediatrician with the diagnosis of microencephaly, spastic diplegia and developmental retardation.

Subsequent care took place near the patient's home with routine medical care by the pediatrician. The child had roseola without seizures. The following developmental milestones were recorded: rolled over at eight months, sat at 12 months, first words at 10 months, and stood with sup-

port at 18 months. Ophthalmologic evaluation was accomplished at 13 months of age when a variable esotropia was observed together with myopic astigmatism in both eyes. He was seen on two occasions at the Santa Rosa Cerebral Palsy Clinic by a pediatrician and orthopedist who outlined exercises for tight heel cords. Arrangements were made for home therapy by an occupational therapist through Atypical Infant Development. With this highly concerned treatment, involving weekly home visits by an occupational therapist, good progress began. The parents were instructed in rehabilitation measures which resulted in unexpected levels of functional recovery.

A full-term 6 lb., 8 oz. female sibling was born February 11, 1972. It was during the conduct of this pregnancy that E.S.'s care was transferred to the family physician. The parents were apprehensive about the possibility of another child with developmental retardation and genetic counseling was obtained. This second child has exhibited entirely normal development from birth.

Arrangements were made for E.S. to be seen by the North Bay Regional Center for the Developmentally Disabled. Funding was obtained for further evaluation and treatment beginning with a pediatric neurological examination. E.S. began walking at two years seven months, and was using isolated words as well as demonstrating echolalia. The child was trying to speak but the sounds were unintelligible. Hearing was normal. A spastic quadriplegia was noted which was relatively mild in the lower extremities. His gait was wide-based with increased pronator tone at both wrists. He held objects with the left hand. Stretch reflexes were overactive, plantar responses were extensor, and sensation was normal. Receptive language testing was excellent. It was felt that his mental age was closer to 24 months than to his chronological age of 34 months.

This child's progress accelerated rapidly with instruction of the mother in an exercise program and games to play. Environmental stimulation was greatly enhanced by the acquisition of glasses to correct the refractive error and by surgical correction of the extraocular muscle imbalance. Psychometric testing was accomplished, revealing a probable functioning IQ of 65. The family physician conducted several office visits with the entire family and two home visits, one in the presence of the representative from North Bay Regional Center. Numerous office visits for other general medical problems involving the family allowed frequent communication concerning the progress of this child and his impact on the family. Nursery school placement was accomplished and an overall plan of management of E.S.'s physical, educational, social and emotional needs was outlined on a long-term basis.

Between three and four years of age, improvement in both E.S.'s gross motor and fine motor function was noted. He became enthusiastic about performing games and exercises. Particularly effective were tricycle-riding and exercises in the swimming pool which had been installed by the family. Speech therapy was initiated because dysarthric speech was E.S.'s principal handicap at that time. Socialization was greatly enhanced by nursery school placement, and the child achieved a significant level of independence with regard to dressing, feeding himself and caring for his own toilet needs.

Conflicting opinions between the clinical psychologist and the pediatric neurologist concerning the patient's progress and prognosis had to be interpreted and mediated by the family physician. At age four, the boy's mental age appeared to be closer to three, but there was optimism that with further control of dysarthria and with language stimulation this gap could be decreased. Evaluations by the nursery school teacher as to school performance allowed a more refined estimate of the patient's progress and prognosis.

The matter of family size prompted consideration of permanent contraception. The parents were seen in a counseling session and long-term expectations were discussed concerning financial demands, unusual educational needs, driving privilege, athletics, possible sexual problems at puberty, the stigma of epilepsy and cerebral palsy and, ultimately, marriage and vocational training. After careful deliberation a vasectomy was performed on the father.

Ongoing evaluation and coordination of care continues at approximately quarterly intervals with meetings of the entire family, usually at the time of a routine medical visit, such as a well-baby examination of the patient's sister. These quarterly visits involve routine examinations to monitor growth and development, update immunizations, review of medications and assessment of the current status of therapeutic modalities. Equally important is discussion of feelings of all the family members concerning the child with this special problem. Short and long-term goals of education are discussed and the child's progress in nursery school is periodically reviewed. Considerable progress has been noted in the social interaction the boy has enjoyed during supervised play among his peers.

Discussion

The above case report illustrates some of the important points that arise in the diagnosis and management of the child with developmental retardation. Initial assessment and re-evaluation of such a child occurs actively in the first years of life while rapid developmental changes, both psy-

chological and physical, are taking place. In this case, such evaluation included a pediatric neurological assessment at three intervals, consultation with an ophthalmologist, otolaryngologist, orthopedist, clinical psychologist, speech therapist, social worker and occupational therapist. A preschool nursery as well as two agencies to assist in funding and coordination of care were also involved. The child's medical evaluation included routine history and physical examination, x-rays, blood tests and two electroencephalograms. The parents have had to spend considerable time over and above what normally would be the case dealing with this particular child's problem. In addition, the danger of conflicting or anxiety-producing comments from such a variety of individuals dealing with this child's problems is potentially great.

It is important that the family physician recognize the great emotional stress on routine family functioning that such a health problem represents. It is essential that the family physician embark on periodic sessions of conjoint family therapy for the purpose of counseling and assessment of disruptive effects on family functioning. In these sessions he should be alert to reactions of denial, guilt and rejection that are commonly present. In addition, the problem of sibling maladjustment, of particular concern in this family, must be dealt with to avoid damaging effects to the sibling(s) during their formative years. A long-term plan should be developed which deals with potential problems such as entry into school, sexual problems at puberty, vocational adjustment, possible marriage and decisions of placement outside the home. Anticipatory discussion should be held with the parents in an effort to prepare the family for these "crises."[6] Much of this counseling can be done at the time of routine medical visits for other family members. It is helpful to encourage the parents to join the local organization of parents with mentally retarded children for the group support such an association affords. The prescribing of drugs such as anticonvulsants and medication for hyperkinetic behavior requires careful discussion because of the likelihood of long-term use and potential side-effects.

Potential pitfalls involved in the care of developmental retardation include the following:[7,8]

1. Conflicting opinions as to diagnosis and prognosis from various professionals.
2. Premature statements with regard to prognosis (slow, careful evaluation is mandatory).
3. Too much emphasis on screening tests and their results, particularly IQ testing.
4. Physician not maintaining professional objectivity and becoming drawn into the emotional process of the family.
5. Failure to recognize the possibility of parental antagonism toward the physician who may become the object of projected hostility resulting from this problem.
6. Excessive re-evaluations, consultations, special studies and unnecessary or protracted treatments of limited value.
7. Failure to dispell misconceptions concerning diagnostic medical labels used to describe the child's deficiencies.

The family physician who is alert to these pitfalls and takes a concerned and open approach to the family presenting with developmental retardation can help them cope more effectively with this challenging but treatable problem. An effort was made to educate this family to a clear understanding of the patient's problem. Parents are interested in specific information regarding their child's current status and future prognosis, as well as the demands likely to be placed on them to meet his needs. In this case, it was possible to dispel the misconception that spasticity and dysarthria signify mental retardation. The concept was explained that compensatory functioning of the alternate cerebral hemisphere occurs in young children with resultant enhancement of ultimate prognosis. The parents were able to verbalize their anxiety with regard to future pregnancies, and emotional and financial stresses as well as the effect on their daughter. As is often the case, the stress presented to the family by this major problem has become a unifying and strengthening influence.

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