Mental Retardation in Tuberous Sclerosis Complex

Early research originally identified the triad of mental retardation, intractable epilepsy, and facial angiofibromas as the trademarks of tuberous sclerosis complex (TSC). We now understand that only about 30 percent to 40 percent of affected individuals have all three features. While many people with TSC have normal intelligence, a large number do in fact suffer from cognitive impairment, or mental retardation.

Mental retardation strictly refers to a person whose intelligence quotient (IQ) is below the normal range, or less than 70. A deficit in adaptive behavior must also be present. This involves problems coping with new or different situations in a classroom or everyday life. When a person cannot cooperate with standardized IQ testing, such as is the case with young children and infants or the severely impaired, a psychologist can perform a standardized rating or estimate of cognitive ability based on observations of the person.

After 8 years of age, **and** in the absence of confounding factors, an IQ or other rating is generally felt to be stable over time. Some things that can affect an IQ rating include medications, ongoing seizure activity, whether a person is trying his or her best, is anxious, frightened or tired, etc. Whoever performs the test or evaluation must find out enough about the person being tested to determine if factors like these are affecting the result.

Diagnosis

It is critically important that the diagnosis of mental retardation be based on objective criteria applied by qualified people, such as a psychologist or psychiatrist. Often caregivers, therapists, teachers, relatives, acquaintances, and even physicians may say that someone is "retarded" based only on their opinions or subjective impressions. While no harm may be intended, this kind of imputation can become a "label" that sticks with someone for many years and is never questioned or re-evaluated. This is particularly true for children younger than 8 years of age, where assessments largely involve rating of intelligence by another person. The harm is that the person may not receive needed evaluations or services that could minimize deficits and improve function. For example, autism is often confused with mental retardation. While the two conditions can co-exist in the same person, many times an autistic person in fact has normal intelligence. A child may be diagnosed with mental retardation while taking sedating drugs or experiencing frequent seizures. If these conditions improve or remit, then significant increases in learning and cognition can occur over time.

Incidence

How common is mental retardation in TSC? The answer is not clear. Doctors used to think that everyone with TSC was retarded. Estimates range from 38 percent to 80 percent in different studies. Currently the best data available suggests an overall incidence of mental retardation of approximately 40 percent in TSC.

A major finding in recent years has been the understanding that the more tubers a person has in their brain, the more likely he or she is to be mentally retarded. While this is true as a statistical association, it does not allow prediction of a person's IQ based on the number of tubers present. Neither is there a "threshold" or safe number of tubers to have, below which a child will not be mentally retarded. It makes sense that the more involvement there is in a person's brain the more likely problems will occur. However, many, a few, or even one tuber can seriously

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disrupt brain function and development in a particular person. Whether or not someone has TSC, we cannot change the genetic "hand" we have been dealt. There are nonetheless many things that can minimize the challenges imposed by mental retardation in this context.

Management

Interventions for mental retardation ideally mean a pro-active approach. Pro-active strategies involve the early identification and treatment of mental retardation, and, whenever possible, the prevention of factors known to aggravate it. Any child who is known to have TSC should be considered at developmental risk, even if there are no signs of abnormality at the present time. For children diagnosed in infancy, referral is made for evaluation and follow-up by a local Early Intervention specialist, who is available through each county's board of mental retardation/developmental disability. A pediatrician or neurologist should closely monitor the child's developmental status at periodic follow-up visits. From infancy to early childhood these should take place a minimum of every three to six months.

In addition to physician assessment, performance of a developmental evaluation, such as the Bayley Scales of Infant Development, is recommended between age 12 to 18 months, and again at 2 to 3 years of age. Less sensitive screening measures used by pediatricians for the general population, such as the Denver Developmental Screening Test, can miss subtle deficits. Any suggestion of difficulty indicates the need for occupational, physical and/or speech therapy. Many physicians assume incorrectly that children cannot benefit from therapy before age 3. Early identification and treatment are critical to help avoid future problems. This is followed by a formal neuropsychologic evaluation at or just prior to entering primary school/first grade. Children who have no evidence of mental retardation at this point are allowed to continue in regular school with standard assessments by their local school personnel. Children with identified cognitive or motor skill problems need to continue/supplement their therapy programs with school-based programs. An individualized educational plan (IEP) should be formulated with particular attention paid to the results of neuropsychologic testing and close involvement of the neuropsychologist. Follow-up neuropsychologic testing with appropriate adjustments in the IEP should be undertaken every two to three years.

Seizures and Mental Retardation

Epilepsy often begins in early childhood/infancy. This is the major treatable factor in TSC responsible for aggravation of cognitive and behavioral difficulties. People with TSC who have the onset of seizures after age 5 are much less likely to have mental or cognitive problems. It is not known if people who are more severely affected with TSC are more likely to have both earlier onset of epilepsy and mental retardation, or if the early onset of epilepsy favors the development of mental retardation. Seizures, as well as seizure activity recorded on an electroencephalogram (EEG), are associated with disruption of brain function and can directly interfere with cognition, learning and language development. These effects can be noticed in the short term, for example, while the person is experiencing the seizures or seizure activity. It can also affect learning over the longer term, if seizures remain uncontrolled for months or years.

Epilepsy Treatment

It is important to provide the best seizure control possible for many reasons. Avoiding aggravation of mental retardation is one of the most important. At the same time, anticonvulsant drugs can often have sedating or "dulling" effects that can themselves hamper learning and cognition. Some anticonvulsants can cause hyperactivity and attention problems. These side effects are more common when multiple anticonvulsants are used. Drugs that have sedative properties such as barbiturates (e.g. phenobarbital) and benzodiazepines (e.g. diazepam) are especially likely to have this effect. Many times any negative effect of medication is more than outweighed by the benefit on cognition, improved seizure control or seizure freedom.

Other Therapies

Finally, many people with mental retardation have other behavioral issues that may be more disabling than their cognitive problems. Hyperactivity, inattention, aggressive behaviors, obsessions or compulsions can all cause major difficulties and secondarily inhibit a person's ability to learn or participate in family/social activities. Behavior modification techniques are very useful for these conditions and are employed first. This involves rewarding or encouraging desired behaviors and discouraging undesirable ones. Consistent application between parents and other caregivers is very important. Guidance from an experienced psychologist or therapist is necessary to implement and monitor such a program. In selected cases, specific medications may be useful as well. Stimulants such as methylphenidate (Ritalin®) or dextroamphetamine (Dexedrine®) can improve attention and a person's ability to focus on assignments. Other drugs can help if learning is impaired by specific behaviors, such as obsessions, compulsions, outbursts of anger, etc. In each case, the possible benefits of drug treatment must be carefully weighed against their side effects in a specific child or individual.

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Made possible through an educational grant from the Schnurmacher Foundations. June 2006