Fact Sheet:
Rett’s Syndrome

Rett’s syndrome (F84.2) is a condition of unknown cause, so far reported only in girls, which has been differentiated on the basis of a characteristic onset, course, and pattern of symptomology. Typically, apparently normal or near normal early development is followed by partial or complete loss of acquired hand skills and speech, together with deceleration in head growth, usually with an onset between 7 and 24 month of age. Hand-wringing stereotypies, hyperventilation, and loss of purposive hand movements are particularly characteristic. Social and play development are arrested in the first 2 or 3 years, but social interest tends to be maintained. During middle childhood, trunk ataxia and apraxia, associated with scoliosis or kyphoscoliosis tend to develop and sometimes there are choreoathetoid movements. Severe mental handicap invariably results. Fits frequently develop during early or middle childhood.

Diagnostic guidelines

In most cases onset is between 7 and 24 months of age. The most characteristic feature is a loss of purposive hand movements and acquired fine motor manipulative skills. This is accompanied by loss, partial loss or lack of development of language; distinctive stereotyped tortuous wringing or “hand-washing” movements, with the arms flexed in front of the chest or chin; stereotypic wetting of the hands with saliva; lack of proper chewing of food; often episodes of hyperventilation; almost always a failure to gain bowel and bladder control; often excessive drooling and protrusion of the tongue; and a loss of social engagement. Typically, the children retain a kind of “social smile”, looking at or “through” people, but not interacting socially with them in early childhood (although social interaction often develops later). The stance and gait tend to become broad-based, the muscles are hypotonic, trunk movements usually become poorly coordinated, and scoliosis or kyphoscoliosis usually develops. Spinal atrophies, with severe motor disability, develop in adolescence or adulthood in about half the cases. Later, rigid spasticity may become manifest, and is usually more pronounced in the lower than in the upper limbs. Epileptic fits, usually involving some type of minor attack, and with an onset generally before the age of 8 years, occur in the majority of cases. In contrast to autism, both deliberate self-injury and complex stereotyped preoccupations or routines are rare.

Differential diagnosis. Initially, Rett’s syndrome is differentiated primarily on the basis of the lack of purposive hands movements, deceleration of head growth, ataxia, stereotypic “hand-washing” movements, and lack of proper chewing. The course of the disorder, in terms of progressive motor deterioration, confirms the diagnosis. Information from: The ICD-10 Classification of Mental and Behavioral Disorders – Clinical descriptions and diagnostic guidelines™ and the following is amended from those pages.