Angelman Syndrome

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Angelman Syndrome Information Page.

Angelman syndrome (AS) is a genetic disorder that causes neurological and psychological problems including seizures, difficult behaviors, movement disorders, and sleep problems. Gastrointestinal, orthopedic, and eye problems also are often present. Infants with AS appear normal at birth but often have feeding problems in the first months of life and exhibit noticeable developmental delays by 6-12 months. Seizures often begin between 2-3 years of age and occur in 80-85 percent of those with AS. Features that help define the syndrome include very happy demeanor with frequent laughter, poor balance, tremor, and minimal to no speech. The disorder results from the absence of the UBE3A gene inherited from the mother. The gene provides instructions for a protein that plays a critical role in the normal development and function of the nervous system.

There are four types of Angelman syndrome involving problems with chromosomes or mutations in the UBE3A gene. Other children may have a genetic syndrome that looks like AS but is caused by a different gene. Dr. Harry Angelman first reported the syndrome in 1965, when he described three children in his practice with similar symptoms.