Facts About AS

What is Angelman Syndrome

Dr. Harry Angelman (1915-1996) and his wife, Audrey (1936-1999)

Harry and Audrey attended several ASF meetings and Audrey corresponded with many US families.

In 1965, Dr. Harry Angelman, an English physician, first described three children with characteristics now known as the Angelman syndrome (AS). He noted that all had a stiff, jerky gait, absent speech, excessive laughter and seizures. Other cases were eventually published but the condition was considered to be extremely rare at that time, and many physicians doubted its existence. The first reports from North America appeared in the early 1980s. Dr. Angelman relates the following regarding his discovery of this syndrome.

""The history of medicine is full of interesting stories about the discovery of illnesses. The saga of Angelman''s syndrome is one such story. It was purely by chance that nearly thirty years ago (e.g., circa 1964) three handicapped children were admitted at various times to my children''s ward in England. They had a variety of disabilities and although at first sight they seemed to be suffering from different conditions I felt that there was a common cause for their illness. The diagnosis was purely a clinical one because in spite of technical investigations which today are more refined I was unable to establish scientific proof that the three children all had the same handicap. In view of this I hesitated to write about them in the medical journals. However, when on holiday in Italy I happened to see an oil painting in the Castelvecchio museum in Verona called . . . A Boy with a Puppet. The boy''s laughing face and the fact that my patients exhibited jerky movements gave me the idea of writing an article about the three children with a title of Puppet Children. It was not a name that pleased all parents but it served as a means of combining the three little patients into a single group. Later the name was changed to Angelman syndrome. This article was published in 1965 and after some initial interest lay almost forgotten until the early eighties.""

Source: 7th edition Facts about Angelman Syndrome by Charles A. Williams, M.D., Sarika U. Peters, Ph.D., Stephen N. Calculator, Ph.D. in 2009

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In 1987, Ellen Magenis, a physician at the Oregon Health Science Center, identified children with microdeletions of chromosome 15 who were expected to have the Prader-Willi syndrome. However, these children had seizures and severe developmental delay, features not expected to be found for that syndrome. It was quickly realized that these children had microdeletions on the maternally derived number 15 chromosome whereas in the Prader-Willi syndrome the deletion was always observed on the paternally derived one. This was an important discovery and ultimately paved the way for the delineation of several mechanisms that caused AS, all by disruption of a gene located on chromosome 15. It was learned that the syndrome can be caused by two copies of the paternal chromosome 15 (1991) and that a regulatory region (the Imprinting Center) can be also be disrupted to the syndrome (1993). In 1997, 10 years after the chromosome deletion was identified, the AS gene, UBE3A, was isolated. This discovery quickly led to the development of animal models and to active neuroscience research aimed at discovering how abnormalities of UBE3A cause impairment in neural development.

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