Aicardi-Goutieres syndrome, a rare neurological disease in children: a new autoimmune disorder?


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Abstract

Aicardi-Goutieres syndrome (AGS), described by J. Aicardi and F. Goutieres in 1984, is a rare neurological disease with onset in infancy. It is often misdiagnosed as a sequela of congenital infection or recognized later. Nowadays almost 200 cases are reported all over the world, most of them collected by the International Aicardi-Goutieres Syndrome Association (IAGSA), founded in Pavia (Italy) in 2000. AGS (MIM 225750) is a genetically-determined encephalopathy characterized by severe neurological dysfunction, acquired microcephaly associated with severe prognosis quoad valetudinem, and less frequently also quoad vitam. Some AGS children also develop some symptoms overlapping with systemic lupus erythematosus (SLE). Intracranial calcification, white matter involvement and brain atrophy revealed on MRI, lymphocytosis and elevated levels of interferon alpha (IFN-α) in the cerebrospinal fluid (CSF) are features of both AGS and congenital viral infection. No evidence of congenital infection at serological exams has ever been found. A genetic etiology was hypothesized since the first descriptions, because of the recurrence in families, and demonstrated some years ago. Nowadays five genes (AGS1-5), if mutated, can be responsible for 90% of the cases. The transmission is autosomal recessive but there are also rare “de novo” autosomal dominant cases. Even if pathogenesis is still almost unknown, it seems that responsible genes are involved in nucleic acid reparation mechanisms and consequently in a secondary activation of innate autoimmunity. The relative lack of precise information on pathogenesis and on the evolution of the disease over time has not yet allowed the creation of codified diagnostic and therapeutic models and programs.

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