Specific clinical and evolutionary aspects of adenosine deaminase 2 deficiency (DADA2)

Adenosine Deaminase 2 deficiency is an auto inflammatory disease related to a mutation in the CECR1 gene encoding the ADA2 enzyme. Clinically there are three main types of manifestations: vascular, neurological and hematological. We report a series of cases of DADA2 by highlighting the clinical and evolutionary peculiarities. The patients are from the same family resulting from a 2nd degree consanguineous marriage.

The diagnosis is confirmed for the first time in the youngest child H A born in 2007 following a recurrent inflammatory syndrome. The CECR1 mutation was c. [139G> A]; [139G> A] p [Gly47Arg]; [Gly47Arg]. this patient has a micro crania.

The second case was born in 1998 with recurrent fever, since the age of 2 years, hypertension since the age of 6 years. In March 2016 the patient had a febrile seizure with headache, dizziness and abdominal pain, MRI exploration found an appearance of cerebral vacuities in May 2016, a sudden drop in visual acuity, and MRI had diagnosed retro bulbar optic neuritis. This patient has the same mutation as his brother. Currently stable under corticosteroid outside Optic Neuritis.

The third, born in 1986 with edemas of the feet and hands with egg allergy, treated as a Still’s disease for several years now in remission.

Other two cases born in 1984 and 1994, the two died at the age of 17 years, by chronic renal failure for the first and chronic macroscopic hematuria for the 2nd.

The DADA2 disease is pathology with multiple clinical expression, the evolution is sometimes fatal

Biography

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