

Editorial

Acquired microcephaly in a child with absent speech: what are the first line exams?

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Microcephaly is defined as reduce of the head circumference less than 2 SDS for age, sex and gestation. Most situations are congenital, but in some cases the head circumference reduces some months after birth. When acquired microcephaly is associated with absent speech, there are some clinical signs that can help us to achieve the final diagnosis.

1 - acquired microcephaly after 6-12 months, absent speech, delay motor skills, seizures, movement disorder with ataxia and/or limbs tremor, frequent laugh, wings movements, fascination for water, sleep disorder, we need to study the UBE3A gene. There are four molecular defects of this gene: deletion of maternal chromosome 15q11-q13, responsible for 70 % of cases, paternal uniparental disomy, where both copies of chromosome 15 are inherited from de father, imprinting center defects and finally point mutation in UBE3A



Angelman Syndrome

2 - acquired microcephaly since 12 months of age, absent speech, mental retardation, behavioral problems in boys and learning disabilities in girls. In this case, the next step is confirmed of high levels of creatine in urine, increased creatine/creatinine ratio, absence of the creatine signal in magnetic resonance spectroscopy of the brain and confirmation of positive SLC6A8 gene



Creatine Transport Deficiency

3 - acquired microcephaly since the first year of age, absent speech, delay motor skills, hypotonia, cardiomyopathy, liver dysfunction, FreeT4 low, growth hormone deficiency, insipidus diabetes, cerebral magnetic resonance imaging of the brain showing global loss of volume of white matter or thin corpus calosum, lactic acid, alanine and glycine level high. We need to do molecular and biochemical study, muscle/skin biopsy. to confirm the diagnosis



Respiratory Chain Disorder

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