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INHERITED AUTOINFLAMMATORY ENCEPHALOPATHY IN THE DIFFERENTIAL DIAGNOSIS OF CONATAL VIRAL INFECTIONS- NEWBORN WITH AICARDI-GOUTIÈRES SYNDROME

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Aicardi-Goutières (AGS) syndrome is a rare early-onset subacute encephalopathy, usually inherited by autosomal recessive trait. The disease commonly manifests in the first year of life. It is genetically heterogeneous disorder associated with mutations of seven different genes. The pathogenic mechanism is disturbed metabolism of endogenously produced nucleic acids and type I interferon upregulation that activates an immune system response. The main clinical features are encephalopathy, irritability, seizures, truncal hypotonia, peripheral spasticity, dystonic posturing, microcephaly, and intellectual impairment. Some patients have episodes of aseptic febrile illness and chilblain-like skin lesions on fingers and earlobes. In 20% of patients, clinical presentation mimics congenital viral infection (hepatosplenomegaly, elevated aminotransferases, thrombocytopenia); for which the syndrome was previously described as pseudo-TORCH. Characteristic findings on neuroimaging are basal ganglia calcifications, cystic leukodystrophy, brain atrophy, and lymphocytosis in the cerebrospinal fluid (CSF). The disease is progressive with lethal outcome during early childhood in 40% of the cases. No etiological treatment is available, but there are ongoing clinical trials with antiretroviral and immunomodulatory agents. The objective of this case report is to increase awareness of AGS in the differential diagnosis of early-onset encephalopathies with basal ganglia calcifications and leukodystrophy and in newborns with suspected congenital viral infections.

Case Presentation The male patient is the first child of non-consanguineous healthy parents. Delivery was on term, but newborn was small for gestational age (BW 2320 g, BL 46 cm, HC 31 cm), and also had truncal hypotonia, tremor, and splenomegaly. Laboratory results showed thrombocytopenia and pleocytosis in the CSF. Brain ultrasound showed ventriculomegaly and periventricular and basal ganglia calcifications. PCR and serology tests for viral infections were negative. Brain MRI revealed polymicrogyria, leukodystrophy, parenchymal and subependymal calcifications, ventriculomegaly, and cerebellar hypoplasia. AGS was suspected and the diagnosis was confirmed by genetic test. Patient was homozygous for common pathogenic mutation c.314G>A (p.Arg114His) in the TREX1 gene. In the further course, the patient was irritable and encephalopathic, with nystagmus, tremor, hyperkinesia, hypotonia, microcephaly, and severe psychomotor retardation.

Conclusion AGS is a rare autoinflammatory encephalopathy with a broad phenotypic spectrum, including perinatal-onset disease that mimics congenital viral infection. Early clinical suspicion followed by genetic testing can shorten the diagnostic odyssey. The correct diagnosis is required to provide information about the expected course of the disease, genetic counseling of the family, and to offer prenatal diagnosis in subsequent pregnancies.

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EMANUEL SYNDROME – CASE REPORT OF A RARE UNBALANCED TRANSLOCATION SYNDROME

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Emanuel syndrome is a rare syndrome characterized by prenatal and postnatal growth retardation, hypotonia, severe mental retardation, microcephaly with characteristic facial features including micrognathia, protruding forehead, broad and flattened nasal root, long and pronounced philtrum, ear deformities with preauricular pits and/or appendages, high or cleft palate and congenital heart defects, kidney anomalies and genital anomalies in men. It is an unbalanced translocation syndrome caused by a 3:1 meiosis I malsegregation during gametogenesis in a phenotypically normal, balanced translocation carrier parent. The exact incidence of the syndrome is unknown but it is estimated that there are about 500 patients worldwide with this syndrome. We report a male newborn from the mother's 4th pregnancy and 1st birth in which a diagnostic process, done due to intrauterine growth retardation, raised a suspicion of Dandy Walker malformation. After birth, specific characteristics were noticed that raised suspicion of Emanuel syndrome. The diagnosis of Emanuel syndrome is made by detecting duplication 22q10-22q11 and duplication 11q23-qter on the supernumerary derivative chromosome 22 [der (22)], so extensive diagnostics was performed on our patient, including genetic analysis. Genetic analysis confirmed the karyotype 47, XY, + der (22) t (11; 22) (q23, q11.2) mat; CMA and FISH confirmed that it indeed was Emanuel syndrome. Cytogenetic analysis of family members revealed that the mother was a carrier of a balanced translocation between the long arms of 11th and 22nd chromosome that she inherited from her father, and we found the same translocation in one of the mother's sisters. The outcomes of future pregnancies of a parent who is a carrier of balanced translocation t(11; 22) are varying from the possibility of having a healthy child, a child who has Emanuel syndrome, a child who is a carrier of balanced translocation t(11; 22), but also having repeated spontaneous miscarriages. Prenatal diagnosis in these cases is highly recommended. It is necessary to make a diagnosis as soon as possible in order to improve the quality of life and prolong the survival of the patient but also to give timely genetic counselling advice to family members.

Metabolic Disease

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ENT CONDITIONS AND DISORDERS IN CHILDREN WITH HUNTER SYNDROME

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Objective to describe the features of the ENT conditions and disorders in children with Hunter syndrome. Hunter syndrome, also known as mucopolysaccharidosis type II (MPS II),