Aicardi–Goutières (AGS) syndrome is a rare early-onset subcute encephalopathy, usually inherited by autosomal recessive trait. The disease commonly manifests in the first year of life. It is genetically heterogenous 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 childlike-like skin lesions on fingers and earlobes. In 20% of patients, clinical presentation mimics conatal 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 conatal 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 a 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 conatal 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.

Metabolic Disease

**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),