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Introduction: The evolution of genetics is a continuous, dynamic process, so the identification of some monogenic causes of etiologies for severe forms of epilepsy is a topic of current interest. The SPATA5 gene was recently associated with neurodegenerative afflictions, suggesting it plays an important part in the neuronal development. The phenotype associated with the SPATA5 variants includes intellectual disability, drug resistant epilepsy, microcephaly and hearing loss in childhood.

Methods: We are presenting the case of a male patient, aged 3 years and 11 months, which presented for the first time in our clinic at age 4 months, with seizures with recent onset (3 months of age), with mild motor developmental delay, bilateral congenital sensorineural hearing loss and some particular features.

Results: Through thorough investigations which were necessary to determine the etiology in this particular case (structural, metabolic, genetic), WES genetic testing revealed a variant in the SPATA5 gene, variant that explained this patient’s phenotype.

Conclusions: SPATA5 variants associate a syndrome characterized by severe global developmental delay, drug resistant epilepsy, hearing loss and microcephaly, meaning that genetic counseling and testing are of utmost importance in patients with epilepsy, not only in the medical approach to the case, but in the family's overview of the affliction as well.

Key words: SPATA5, genetics, drug resistant epilepsy, WES, global developmental delay