RETT SYNDROME DESCRIBED IN A CHILD AT A CLINICAL FOLLOW-UP: A CASE REPORT

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Rett Syndrome is a neurodevelopmental disorder that mainly affects females. It is a disorder characterized by adequate development during the first 6 months of life, followed by a regression of the neuropsychomotor development. This case report aims to describe a clinical case report of this rare syndrome in a 5-year-old child. The patient's parents went to Associação de Pais e Amigos dos Excepcionais (APAE) reporting loss of head support at 7 months, loss of ability to hold objects, delays in child development stages and seizures. After a neurological appointment, it was supposed that this was a typical case of this rare syndrome. The patient attended APAE from 3 to 5 years old. Currently, this follow-up has been interrupted without explanation. Thus, the reported case and published studies about Rett Syndrome clarify the discussion about the clinical and genetic diagnosis, in addition to the pathophysiology, particularities and prognosis of the syndrome. The diagnosis of the disease is clinical, but genetic testing of the MECP2 gene is requested for confirmation. The severity of symptoms in this rare syndrome are related to mutations in this gene. This syndrome can be confused with Autism Spectrum Disorder, but the loss of developmental milestones are key for distinguishing both disorders.

Keywords: neurodevelopmental disorder; Rett Syndrome; MECP2.

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