DOI: 10.5281/zenodo.4958770

CONFERENCE ABSTRACT

Rett Syndrome- A case report

Sofia Pimenta*, Margarida Peixoto, Margarida Reis Lima, Natália Salgueiro, Marta Vila Real, & Fátima Santos
*Child and Adolescent Neurosciences Unit, Paediatrics Department, Centro Hospitalar Vila Nova de Gaia/Espinho, Portugal

Introduction

Rett syndrome (RTT) is a progressive neurodevelopmental disorder, the most frequent cause of mental retardation in girls. The majority of RTT is caused by mutations in the MECP2 gene, although there are numerous genes that can be involved. Infants with RTT generally develop normally for about 7 to 18 months after birth. At this point, they lose previously acquired skills, such as purposeful hand movements and the ability to speak.

Case report

We present a case of a 20-month-old girl that was referenced to Neurodevelopmental consultation due to a developmental delay. She was the second daughter of healthy non-consanguineous parents. The family history was unremarkable. During the first months of life, there were no significant developmental problems. During the second year of life, she presented a language delay followed by a development regression accompanied by a deceleration of head circumference and stereotypic midline hand movements. Physical examination was unremarkable.

Rett syndrome was suspected and cerebral MRI, low resolution micro-array, metabolic study, karyotype were normal. Fragile X syndrome and Angelman syndrome were excluded. The most common genes of Rett Syndrome (CDKL5, FOXG1, IQSEC2, TCF4, KCNA2, MBD5 e MECP2) previously performed were normal.

At this moment, she is 12 years old and maintains a significant deficit in social interaction and communication, self-aggressive behavior, midline stereotypies, characteristic hyperventilation episodes and epilepsy partially controlled with antiepileptic drugs.

Whole exome sequencing with CNV analysis revealed a 711 bp deletion, involving exon 4 of the MECP2 gene. This deletion is not available in the literature consulted, however, there are several published studies with deletions in this exon associated with Rett syndrome.

Discussion

Rett syndrome is a clinical diagnosis that requires an early multidisciplinary approach. If the initial genetic study doesn´t reveal any anomalies, an extended study should be performed, since the identification of the genetic mutation involved is important for better characterization of the disease and genetic counseling.

Keywords: Rett syndrome; development regression;

**Presenting Author*

Sofia Pimenta. Completed her Integrated Master in Medicine in 2015 at Faculty of Medicine of University of Porto (Portugal). In 2017 she started the Pediatric Residency at Centro Hospitalar do Tâmega e Sousa and currently she works at this hospital.