Our experience in genetic diagnosis of Rett syndrome by Next Generation Sequencing

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Objective:

In this project we present the genetic study of patients RTT-like using Next Generation Sequencing (NGS).

Material and Method:

We have designed a panel of 17 genes related to the RTT-like clinic by *Target HaloPlex technology. Enrichment System, for Illumina Sequencing*, completing the study with Sanger sequencing in regions that are not well covered and performing MLPA major genes causing RTT (Salsa probe: P015, P189 and PXX).
We studied patients with clinical RTT without genetic diagnosis as well patients with negative genetic study of RTT by Sanger and MLPA.
NGS results have been verified by Sanger sequencing and studied the origin of the mutation in the parents.

Results:

It have been detected mutations in the genes that cause RTT as well as other genes related to RTT-like.

Conclusions:

The genetic study by NGS allows us to study a higher number of genes associated with RTT simultaneously, reducing significantly the time response and cost of the study. It also allows us to study other genes related to clinical RTT and thus to redirect the clinical diagnosis to another pathology.
Verification by Sanger sequencing of the mutations detected by NGS in the progenitors and patient remains essential for characterization as well as the need for functional studies.

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