

Temple syndrome as a differential diagnosis in Chilean population with suspected Silver Russell syndrome

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Background: Temple Syndrome (TS14) is a rare imprinting disorder secondary to alterations at the 14q32 imprinted locus. One of its main differential diagnoses is Silver Russell Syndrome (SRS) caused by aberrations of the imprinted 11p15 region or by maternal Uniparental Disomy of chromosome 7 (matUPD7) in most cases. Both TS14 and SRS share within their phenotypes delayed growth associated with feeding difficulties during childhood.

Objective: To identify patients with TS14 in individuals with a negative molecular study result of the imprinted region of chromosome 11p15 with clinical suspicion of SRS.

Methods: We studied 20 individuals with a negative result of MS-MLPA BWS / SRS with clinical suspicion of SRS, previously analyzed at the INTA Molecular Cytogenetics laboratory between the years 2017 and 2021, using MS-MLPA UPD7-UPD14.

Results: The diagnosis of TS14 was confirmed in 1 individual (5%) with 0% methylation in the three probes that analyze the imprinted 14q32 locus, without alteration in copy number in the region. This result is compatible with an epimutation or matUPD14 at the locus. No other molecular alterations were identified with this MS-MLPA salsa in the rest of the studied individuals.

Conclusion: TS14 is probably underdiagnosed, and the molecular study of the imprinted region 14q32 in patients with suspected SRS with a normal study of the imprinted region 11p15 should be considered in the diagnostic algorithm, given their phenotypic overlap.

Conflict of interest disclosure: The authors declare no potential conflicts of interest, whether scientific, financial and personal.

Keywords: Temple syndrome, Silver Russell syndrome, MS-MLPA, 14q32, UPD14