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ABSTRACT BOOK

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Robin sequence, atypical face and intellectual disability in brazilian siblings

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Background: The Pierre Robin sequence (RS) is a rare heterogeneous condition, characterized by the combination of micrognathia and glossoptosis, which can cause respiratory obstruction and severe eating difficulties. Palate cleft, variable feature and has often been incorporated into the definition. It occurs in isolation or associated with other anomalies, characterizing several syndromic conditions or not in a recognized standard such as a known syndrome or genetic condition, termed Robin Plus sequence (RS-plus). Here, we reported two Brazilian siblings, both male, with RS-plus with moderated intellectual disability and atypical face consisting of frontal hirsutism, low hair implantation, prominent thick eyebrows, narrow eyelid fissures, middle face hypoplasic, long and prominent filter, broad nasal bridge and base, hypoplasic nostrils and collumela, broad lobe ear, cleft palate. They also presented small hands, short digits, plantar hyperkeratosis, developmental delay and learning difficulties. Karyotype was normal. The evaluation of language showed an important oral and written language disorder, indicating cognitive deficit. The parents was phenotypically normal.
Aims: To investigate possible chromosome microdeletion or microduplication related to the phenotype of the siblings.
Methods: Microrearrangement analysis by MLPA in subtelomeric region (P036) and multiple microdeletion syndromes (P064).
Results: No change in the regions specific to syndrome with intellectual disability or subtelomeric region were found.
Summary/Conclusion: The same pattern of clinical findings observed in siblings with RS-plus suggest that this phenotype represent a new syndrome. The absence of changes in the subtelomeric regions, as well as in regions of known microdeletion/duplication syndromes, points to a probable genetic etiology. Additionally, the recurrence in male siblings and the presence of intellectual disability suggest that this is an autosomal recessive or X-linked condition.
Additional molecular studies in collaboration with European group have being planned to clarify the etiology of this rare RS-plus condition.