



TITLE: Rubinstein - Taybi syndrome: phenotypic characteristics and case report.

Name: Josue Rivadeneira-Dueñas^{1,2}, César Vaca- Salazar³

Affiliation: ¹Universidad de la Frontera, ²Zero Biomedical Research, ³Hospital Enrique Garcés

Country: Chile / Ecuador

Email ID: josue.rivadeneira04@hotmail.com

ABSTRACT (up to 300 words)

Introduction: Rubinstein-Taybi syndrome is a pathology of genetic origin that affects 1 out of every 100,000 to 125,000 live births, featuring submitting growth retardation, delay in psychomotor development, and morphological abnormalities, mainly in the facial features (thick arched eyebrows, downward sloping palpebral fissures, the convex nasal bridge with the tip of the nose below wings), and the fingers (widening of thumbs and halluxes). Its epigenetic origin in 60% of cases is due to an alteration in the CREBBP gene, in 10% a change in the EP300 gene, and in 30% there is no identifiable cause.

Clinical case: An 8-year-old boy with a delay in psychomotor development had difficulties in adapting to school. On physical examination, facial features showed overpopulated and arched eyebrows, hirsutism in the forehead and upper lip region, downward sloping palpebral fissures, hypertelorism with convergent strabismus, wide nasal bridge, and flattened nose. The tip extends slightly below the nasal wings, with hirsutism in the cervical and interscapular regions. In the hands, broad thumbs are identified. In the rest of the fingers, there are widened distal phalanges, and in the same way, in the region of the feet, a wide hallux and widened distal phalanges are identified.

The patient was sent to speech, reading, and psychomotor therapy programs. He has not developed pulmonary infections until the close of follow-up 6 months after diagnosis.

Conclusion: In this case report, we discuss the phenotypic alterations of the facial and limb characteristics of a child with Rubinstein-Taybi syndrome, which helped the clinical diagnosis.

BIOGRAPHY (up to 200 words)

Josue Rivadeneira-Dueñas is a general practitioner who graduated from Universidad UTE, Ecuador in 2019, with studies in nutrition and research methodology. In 2022 he started his studies for the Doctor in Medical Sciences degree at Universidad de La Frontera (Temuco, Chile). He has been part of national and international research teams, presenting experience in developing, implementing, and monitoring clinical trials and observational studies, having several publications in indexed journals and oral presentations. For these reasons, since 2021 he is a co-founder and active member of the research group Zero Biomedical Research, located in Quito, Ecuador.



International Conference on
PEDIATRICS AND HEALTHCARE
May 22-23, 2023 | Tokyo, Japan

Presenter Name: Josue Rivadeneira-Dueñas.
Mode of Presentation: Virtual/Poster.
Contact number: + 56943849338

