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Case Report Open Access

Introduction

Hallermann-Strei syndrome (HSS) was rst time described by Hallerman in 1948 and Strei in 1950 who distinguished it from progeria and mandibulofacial dysostosis. Disease causing pathogenic mutation in GJA1 gene has been identi ed in a patient with clinical features that overlap Hallerman-Strei syndrome (HSS; MIM# 234100) with Oculodentodigital dysplasia (ODDD; MIM# 164200) [1] However none of the cases with typical HSS are found to contain mutations in the GJA1 gene. We describe an Indian patient with typical HSS rst time without identi able GJA1 mutations.

Clinical Brief

Seven year old girl of Asian- Indian origin presented to us with complaints of short stature and sparse scalp hair. She is the rst child of non-consanguineous marriage. Mother's and father's age were 23 and 25 years respectively at the time of birth. During antenatal period mother had gestational diabetes mellitus controlled with dietary advice and medications. Proposita was born by caesarian section with birth weight of 2.5 kg (15th centile). She was operated for bilateral eye cataract at one month of age. Her vision is 6/6 both eyes (Snellen visual acuity chart) with appropriate refractive correction. Her parents complained of snoring from neonatal age. She developed obstructive sleep apnea syndrome and was prescribed positive pressure ventilation at nights.

well-characterized patients with HSS. Paznekas [2] and Pizzuti [1] also did molecular analysis of six patients with typical HSS and no changes in the sequences of the GJA1 gene were found. None of the patients including ours diagnosed with HSS have been found to have identi able mutation. ough a possibility of a variation in the non-coding region of the gene that a ects the expression of the gene remains, it is more likely that HSS does not come under GJA1 spectrum of disease phenotype and a di erent gene is responsible for this disease. Typical cases of HSS should have GJA1 gene analysis to further support this. Other candidate genes should be searched for by newer sequencing technology.

Acknowledgement

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References

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