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A Novel Translocation t(2;9)(p23;q13) in Female with Short Stature

Rashmi Talwar Sethi, Sunil Kumar Singh Vats, Mohit Kumar Verma and Rohit Agarwal[1]

SRL Diagnostics, Gurgaon, Haryana, India
[1]Amar Hospital, Patiala, Punjab, India

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ABSTRACT Short stature has, for long time, been a diagnostic dilemma for clinicians and a major concern for females and their parents. Although short stature has been observed in individuals with Turner syndrome (45,X) and its variants, there are very few reports on autosomal anomalies being present in individuals with short stature. We present the first case of a 12 years old female with short stature, slightly elevated FSH (10.08 mIU/ml), small sized uterus and ovaries that revealed a balanced reciprocal translocation between chromosomes 2 and 9 [t(2;9)(p23;q13)]. The present case suggests that genes for human height could be mapped to short arm of chromosome 2 and/or long arm of chromosome 9.