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# Urofacial (Ochoa) Syndrome - A Case Report

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**ABSTRACT** Urofacial (Ochoa) syndrome is a very rare autosomal recessive disorder characterized by a peculiar inverted facial expression, mainly when smiling or crying and urinary abnormalities. A 5-year-old boy, born of non-consanguineous marriage presented with characteristic facial grimace, enuresis, constipation and delayed milestones. An evaluation of GTG–banded metaphases revealed a normal karyotype with a paternally inherited variant chromosome 14 (14ps+). However, his father showed a normal phenotype. This is the first report of Ochoa syndrome from India.