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Oro-Facio-Digital Syndrome Type IX with Polydactyly and Multiple Intraocular Findings

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ABSTRACT Oral-facial-digital syndromes are characterized by abnormalities in the oral cavity, face and digits. To date, 13 types with different modes of inheritance have been distinguished based on characteristic clinical manifestations. The researchers reported a twelve-year-old male patient with the common features of oral, facial and digital abnormalities of an OFDIX syndrome along with some unreported features. On assessment, it was found that he had a short stature and microcephaly. The patient had sparse scalp hair and alopecia areata, which is reported only in females with OFDI. He showed hands postaxial polydactyly and unilateral bifid big toe. The patient displayed extraocular manifestations along with multiple intraocular findings. There were additional CNS findings in the form of subependymal and periventricular andarachinoid tiny cysts. The arachinoid cysts were previously described in OFD I, II and III. This case overlapped with the clinical picture between OFD I, which is detected only in girls and OFD II.