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Oral-Facial-Digital Syndrome Type I

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Oral-facial-digital syndrome (OFD) is a collective term, which includes thirteen distinctive, genetic disorders. These syndromes are a form of ectodermal dysplasia, which affects the development of the skin, nails, hair, eyes, face, fingers, toes and the oral cavity. In addition, cardiovascular, renal and central nervous malformations are commonly associated with these conditions. The affected individuals may present with speech and learning disabilities, impaired mental ability, and seizures which are directly related to the severity of central nervous system involvement.

The inheritance pattern and phenotypic expression mainly distinguish the rare 13-subtypes. OFD type I has the highest incidence compared with the other subtypes, and equally distributed among different races and ethnicities. We present a thirteen-year-old female who presented with OFD syndrome, type I. OFD syndrome is a form of ectodermal dysplasia affecting the development of the skin, nails, hair, eyes, face, fingers, toes and the oral cavity.

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