CLINIPICS

Oro-facial-digital syndrome

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Oro-facial-digital syndrome (OFDS), autosomal recessive condition consists of a group of heterogeneous disorder characterized by abnormalities in oral cavity, face and digits. Associated phenotypic abnormalities that lead to delineation of 13 OFD subtypes with different modes of inheritance. The features of different subtypes overlaps and some types are not yet well defined.[1] Classification system for OFDS continues to evolve as researchers found more affected individuals and learn more about this disease.[2]

OFD type II is characterized by frontal bossing, broad nasal bridge, midline cleft lip and palate, high arch palate, lobulated tongue, syndactly, polydactyl and duplication of first toe. Other systemic features include conductive deafness, congenital cardiac defects, choroidal coloboma and normal intelligence. Patient is 25 yr female with high arch palate, lobulated tongue, syndactly, polydactyl, normal hearing and intelligence and no cardiac and ophthalmic problem.

Figure 1: Showing high arch palate and absent uvula

Figure 2: Showing lobulated tongue

Figure 3: Showing syndactly of both foots

Figure 4: Showing syndactly and polydactyl of both hands

REFERENCES

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