

Mammary-Digital-Nail Syndrome With Orofacial Changes

A 12-year-old premenarchal girl presented with the complaint of progressive increase in size of her breasts for one year. There was no family history of dysmorphism, gigantomastia or onychodystrophy/anonychia. Her general examination showed dysmorphic facies with maxillary hyperplasia, maloccluded/ malaligned, hypoplastic (conical) teeth, and prominent nose with flat nasal bridge, anteverted nares and short columella (**Fig. 1A, 1B**). Bilateral brachydactyly of all fingers except the middle fingers was noted in the form of hypoplasia of distal phalanges. Anonychia was seen in all fingers and toes. The big toe of both feet appeared hypoplastic (**Fig. 1C**).

Her bilateral breasts were grossly enlarged, disproportionate to age (right more than left side) (**Fig. 1D**). A 8×5 cm mobile and non-tender lump was palpable in upper outer quadrant of right breast with no inflammatory changes. Ultrasound of breasts revealed excessive fatty parenchyma, without delineation of fibro-glandular

portion. A well-defined lesion was confirmed in right breast, 9×5.5×8cm size, with benign characteristics.

Her hormonal profile, karyotype and ultrasound of abdomen and pelvis were normal. There was no history of intellectual disability in the girl. A phenotypic diagnosis of Mammary-Digital-Nail syndrome (MDN Syndrome; OMIM 613689) with additional orofacial changes was made. It is mapped to chromosome 22q12.3-q13.1. The patient was referred to surgeons for reduction mammo-plasty and management of benign breast lump.

Other etiologies for breast hypertrophy include fibrocystic disease, juvenile gigantomastia or juvenile hypertrophy of the breast, malignancies and hormonal disorders. Juvenile or virginal hypertrophy of breast (JHB) is a rare clinical entity of undefined etopathogenesis, presenting with gigantomastia in peripubertal girls with no associated abnormality.

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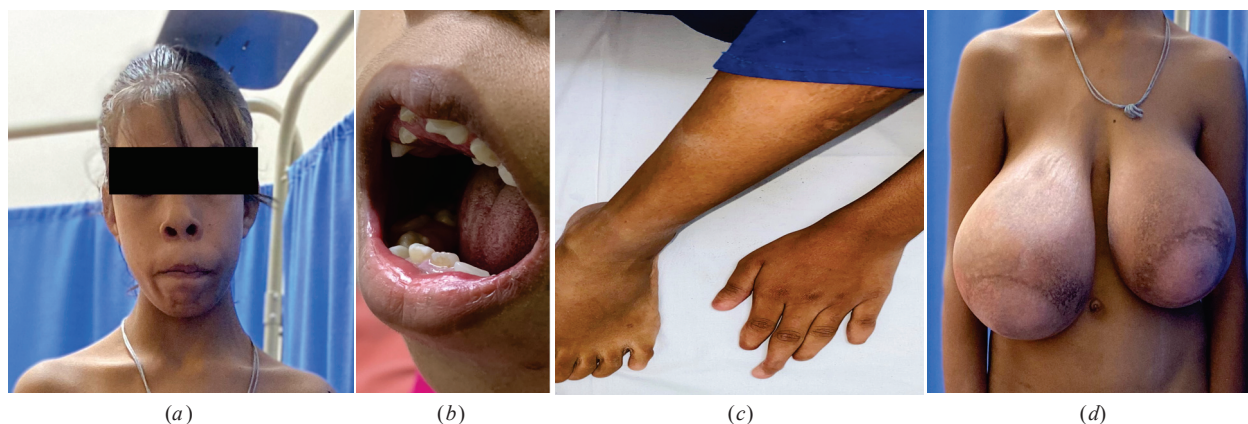


Fig.1 a) Facial dysmorphism, b) dental abnormalities, c) toe abnormalities, and d) breast enlargement in a girl with Mammary-Digital-Nail syndrome.