GENETICS AND GENODERMATOSES

TERMINAL OSSEOUS DYSPLASIA WITH PIGMENTARY DEFECTS IN A CHINESE GIRL WITH FLNA MUTATION

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Background: Terminal osseous dysplasia with pigmentary defects (TODPD) is an extremely rare X-linked dominant syndrome, which is characterized by pigmentary skin defects, cutaneous digital fibromas, and skeletal anomalies. Recent studies revealed TODPD was caused by a unique variant, c.5217G>A (p.Val1724_Thr1739del), in the FLNA gene.

Observation: A 9-month-old Chinese girl was presented in our clinic with multiple dysmorphic features that had been present since her birth. She was the first child of non-consanguineous unaffected parents. Family history is unremarkable. Physical examination showed light brown-to-yellow, slightly atrophy macules in a linear array on the central forehead and bitemporal regions. Other facial features included frontal bossing, blepharoptosis, bilateral epicanthal folds, a broad nasal root and a flat nasal bridge. Future examination revealed dysplastic teeth and an accessory oral frenulum between the right lower lip and right lower gum. Musculoskeletal inspection showed hand and foot contractures, right hand clinodactyly of the first, third, and fifth fingers, left hand clinodactyly of the third, forth, and fifth fingers and left foot clinodactyly of all toes. There was partial soft tissue syndactyly of the second through fourth fingers on her right hand and partial soft tissue syndactyly of third and forth toes on her left foot. Radiological examination showed shortened metacarpals of bilateral thumbs, periostosis and bone destructions in the metacarpals and phalanges. Biopsies taken from the nodules on the lateral aspect of the right third finger were confirmed to be digital fibromas. DNA extracted from her paraffin-embedded tissue was screened for FLNA mutations by complete Sanger sequencing. Genetic test returned positive for FLNA c.5217G>A heterozygous mutation diagnostic for TODPD. Parental test was declined.

Key message: Herein, we reported a rare case of TODPD caused by FLNA mutation which is also the first case reported in Chinese population.