NAIL DISORDERS

CONGENITAL NAIL CLUBBING

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Background: Nail clubbing is characterized by a focal bulbous enlargement of the terminal segments of the fingers and/or toes. It is an isolated abnormality or associated with a systemic disease, showing pulmonary, cardiovascular, gastrointestinal and/or metabolic conditions, as typically seen in primary hypertrophic osteoarthropathy (PHO) / pachydermoperiostosis (PDP). Mutations in the two PHO/PDP-associated genes, HPGD and SLCO2A1, have been reported.

Observation: A 31-year-old man had clubbing nail at birth, noticed oily facial skin and palmoplantar hyperhidrosis at 10-14 years, and developed pachydermia at the age of 20 years. We found remarkable dermal infiltration of mast cells and an increase of urinary metabolite of PG. These findings suggested that he was a case of PHO/PDP, however, we did not find any exome mutations in HPGD and SLCO2A1.

Key message: HPGD and SLCO2A1 have been reported in families of isolated congenital nail clubbing and pachydermoperiostosis. In our case, the patient showed PDP like phenotype and laboratory data, but he had no mutations. Therefore, it is possible that our case is a mild case of PHO/PDP without HPGD or SLCO2A1 mutation.