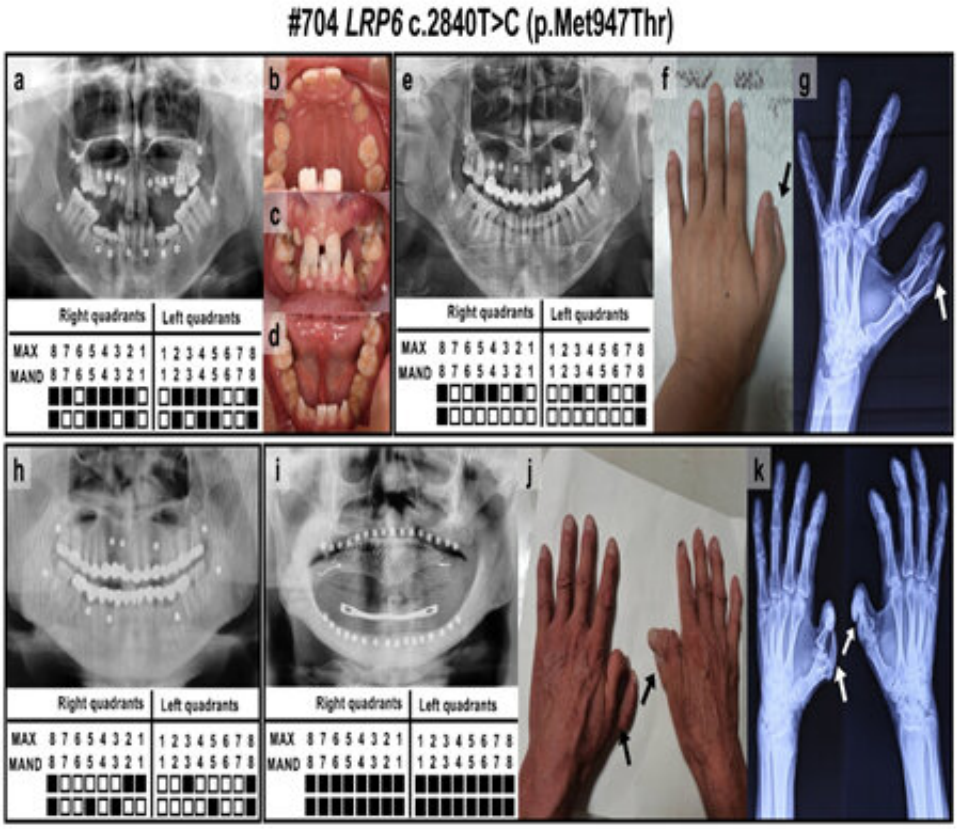
A new phenotype of LRP6 mutation

November 29 2021



Clinical features and pedigree of families with tooth agenesis caused by LRP6 mutations. Credit: Peking University School of Stomatology

A group led by Prof. Han Dong of Peking University School of Stomatology reported for the first time hand preaxial polydactyly in patients with tooth agenesis carrying a previously unknown mutation of low-density lipoprotein receptor-related protein 6 (LRP6). The study was

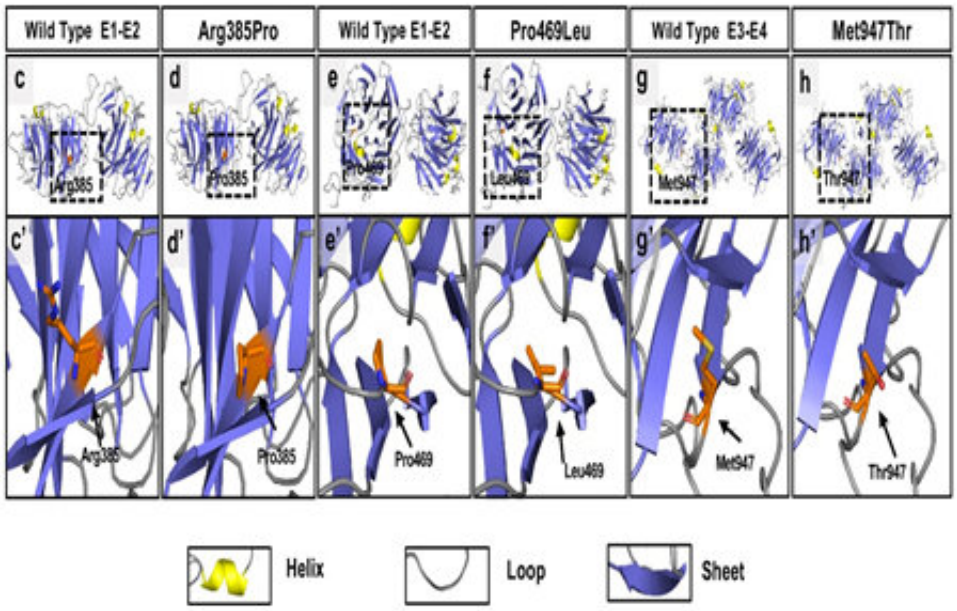
published November 10 in *NPJ Genomic Medicine*.

In the study, the researchers performed whole-exome sequencing and clinical assessment of three families with inherited tooth agenesis to investigate the genetic causes and clinical phenotypes, and reported (1) a rare phenotype of hand preaxial polydactyly in a tooth agenesis family with an unreported autosomal dominant LRP6 heterozygous mutation (c.2840 T > C;p.Met947Thr) and (2) another unreported autosomal dominant LRP6 heterozygous mutation (c.1154 G > C;p.Arg385Pro) in a non-syndromic tooth agenesis family.

The findings enlarged the LRP6 mutation spectrum of tooth agenesis and broadened the phenotypic spectrum of LRP6-related disorders, which may help clinicians to differentiate diagnosis, target pathogenic genes in advance, and facilitate the genetic studies of tooth agenesis patients.

Prof. Han served as corresponding author of the paper, and Dr. Zhang Liutao from the 2021 Class of the school's integrated 8-year MD/Ph.D. program and Assistant Fellow Yu Miao were first authors.

The research group on developmental dental anomalies at Peking University was initiated by Prof. Feng Hailan, and is currently led by Prof. Han. The group has long been devoted to research in the molecular mechanism of developmental dental anomalies and its clinical translation, and boasts the world's biggest biobank and digital information database of tooth agenesis.



Structural modeling of the wild-type and mutated functional domains of LRP6 protein. Credit: Peking University School of Stomatology

More information: Liutao Zhang et al, Rare phenotype: Hand preaxial polydactyly associated with LRP6-related tooth agenesis in humans, *npj Genomic Medicine* (2021). DOI: 10.1038/s41525-021-00262-0

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