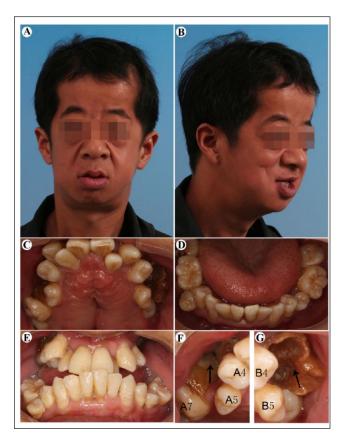
Dental Abnormalities Caused by Novel Compound Heterozygous CTSK Mutations

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Appendix



Appendix Figure 1. Oral and craniofacial manifestations of the patient. The patient displays frontal and parietal bossing, hypoplasia of facial bones, and symmetrical swellings on both sides of the submandibular region in anterior view (**A**) or lateral view (**B**). Intraoral features include grooved palate (**C**), dental crowding in both maxilla (C) and mandible (**D**), and severe malocclusion (**E**). The exposed wounds (arrows) in the maxilla are shown in **F** (the right side) and **G** (the left side).

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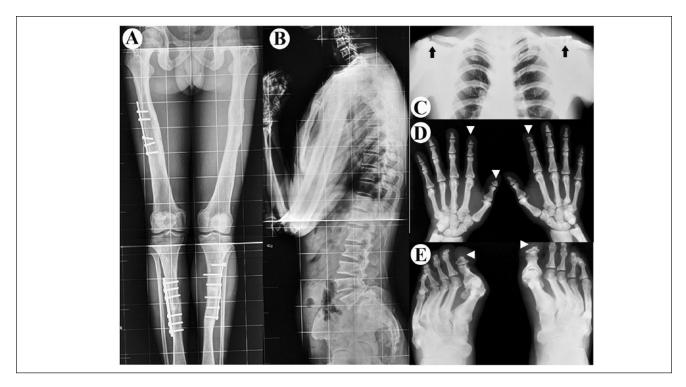
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Appendix Figure 2. Radiologic findings of the patient. Examinations show increased bone density and multiple old fractures in bilateral lower limbs (**A**, **B**), hypoplasia of the clavicles (**C**; arrows), and osteolysis of the distal phalanges in hands (**D**; arrowheads) and feet (**E**; arrowheads).