GENETICS AND GENODERMATOSES

PREMATURE AGING SYNDROME, PENTTINEN TYPE: REPORT OF A CHINESE CASE WITH PDGFRB MUTATION

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Background: Premature aging syndrome, Penttinen type (Penttinen syndrome) is a rare disorder characterized by prematurely aged appearance, delayed bone maturation and dental development, pronounced acro-osteolysis with brachydactyly, distinctive translucent skin with keloid-like lesions and lipoatrophy. Totally, 5 cases have been reported.

Observation: We enrolled 1 Chinese case of Penttinen syndrome. Exome sequencing was performed. The patient presented with characteristic presentations of Penttinen syndrome. Cranial CT and MRI scan showed open anterior fontanel, posterior fontanel and sagittal suture, hydrocephalus, cerebellar atrophy, and leukoencephalopathy which did not match his age. Echocardiography showed left ventricular diastolic dysfunction. Exome sequencing identified a PDGFRB c.1994T>C, p.Val665Ala variant in the patient and Sanger sequencing confirmed this variant as a de novo mutation.

Key message: We confirmed PDGFRB c.1994T>C, p.Val665Ala variant as the causative mutation of Penttinen syndrome. Long-term follow-up is needed to determine the evolution and prognosis of this syndrome.