Commentary

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An Overview on Penttinen-Aula Syndrome

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A rare genetic progeroid syndrome disorder marked by a prematurely aged appearance (including lipoatrophy, thin, translucent skin, sparse, thin hair, and skeletal muscle atrophy), delayed tooth eruption, keloid-like lesions on pressure areas, and skeletal abnormalities including marked acroosteolysis, brachydactyly with small hands and feet, kyphoscoliosis, osteopenia, and progressive joint contractures in the fingers and toes. A small calvarium, delayed anterior fontanel closure, flat occiput, shallow orbits, malar hypoplasia, and a narrow nose are some of the cranial characteristics.

Penttinen syndrome is a rare condition distinguished by lipoatrophy, epidermal and dermal atrophy, hypertrophic scar-like lesions, sparse hair, proptosis, underdeveloped cheekbones, and severe acro-osteolysis. All of the people were simplex instances. An affected person's exome sequencing revealed a *de novo* c.1994T >C p.Val665Ala variation in PDGFRB, which codes for the platelet-derived growth factor receptor. Three more unrelated people with this ailment were discovered to have the same PDGFRB mutation.

Infantile myofibromatosis, idiopathic basal ganglia calcification, and an overgrowth disorder with dysmorphic facies and psychosis have all been linked to PDGFRB mutations, none of which are similar to the clinical features of Penttinen syndrome. Transfecting mutant and wild-type cDNA into HeLa cells revealed ligand-independent constitutive signalling through STAT3 and PLC, indicating that this causal variant has a functional impact on the PDGFRB signalling pathway. Penttinen syndrome is a clinically different genetic disorder caused by a gain-of-function mutation in the PDGFRB gene that results in a distinctive and unusual change in receptor function.

Gain-of-function mutations appear to be the aetiology of both Penttinen syndrome and myofibromatosis. Signal transmission from receptor tyrosine kinases involves a large number of downstream effectors. STAT3 and PLC1 appear to be effectors of the p.Val665Ala PDGFRB Penttinen syndrome mutation, according to our findings. Although PLCG1 mutations have not been linked to disease, STAT3 activating mutations have been linked to cancer and autoimmune disease. Inhibitors of STAT3 phosphorylation in keloid fibroblasts reduce enhanced collagen synthesis, proliferation, and migration, suggesting that STAT3 may play a role in keloid development.

PS stands for progeroid syndromes, a set of rare genetic illnesses that mimic physiological ageing and make people appear older than they are: Progeria (Hutchinson–Gilford progeria syndrome) is a kind of progeroid disease that is not often associated with the word progeroid syndrome. Progeroid is a term that refers to a wide spectrum of disorders that resemble premature ageing. Two well-known accelerated-aging disorders that are more common in older people are familial Alzheimer's disease and familial Parkinson's disease. They are known as unimodal progeroid syndromes because they only affect one tissue. Segmental progeria, which is more commonly referred to as progeroid syndrome, affects several or all tissues while causing affected persons to exhibit only some of the signs and symptoms of ageing.

