GENETICS AND GENODERMATOSES

# CASE REPORT- SIBLINGS WITH ACCELERATED AGEING: PROBABLY THE FIRST OF ITS KIND PROGEROID SYNDROME.

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Background: Progeroid syndrome consist of an overarching group of rare genetic disorders mimicking accelerated physiological aging. They may involve only one tissue, like Alzheimer's, or multiple tissues, like Hutchinson-Gilford Progeria syndrome (HGPS), the most specific type of Progeria. Progeria is an autosomal dominant disorder caused due to a spontaneous mutation in utero. As people with progeria do not reach reproductive age, familial progeria is almost never seen.

Observation: Two, 15 & 10-year-old, short-statured, malnourished brothers presented to us with typical features of progeria including but not limited to prominent eyes, hypoplastic chin, saddle nose, papery thin skin, prominent veins especially over scalp & sparse brittle hair. They gave a history of accelerated ageing. However, both appeared to have age appropriate intelligence. There was no family history of similar complaints.
A radiogram revealed osteopenia, delayed fusion of fontanelles and equinovarus of the feet. Calcified valves were seen in an Electrocardiogram.
Though clinically they appeared to be suffering from HGPS, siblings having progeria has never been reported in literature and it seemed almost impossible that siblings had acquired the same mutation, spontaneously, in-utero.
At a stalemate, we reached out to the Progeria foundation and they confirmed our suspicion. However even they couldn't arrive at an exact diagnosis. Genetic testing was suggested, which confirmed the absence of the mutation causing HGPS. Whole exome sequencing, the next logical step, was unable to reveal any candidate genes despite multiple reads. Currently Whole genome sequencing is being undertaken to ascertain this unique, and probably the first of a kind progeroid syndrome, along with managing their systemic manifestations aiming to extend their life.

Key Message: Despite being autosomal dominant, HGPS is almost never seen in families. Hence, any familial case with progeroid features should be intensively investigated to reach a specific diagnosis.