GENETICS AND GENODERMATOSES

# CASE REPORT: DELAYED DIAGNOSIS OF GORLIN SYNDROME

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Background: Nevoid basal cell carcinoma syndrome (NBCCS) also known as Gorlin Syndrome is a rare autosomal dominant genodermatosis mainly characterized by the presence of a large number of basal cell carcinomas (BCC) at an early age. It is caused by a mutation of the protein Patched homolog 1 gene (PTCH-1), which encodes a key transmembrane receptor within the Sonic Hedgehog (SHH) signaling pathway. The major clinical features are multiple early-onset BCCs, keratocystic odontogenic tumors, palmoplantar pits, falx cerebri calcification and a positive family history. Minor features include skeletal abnormalities, macrocephaly, bilateral ovarian fibroma, medulloblastoma and other congenital malformations.

Observation: A fifty-year-old Panamanian male, Fitzpatrick type IV, was referred to dermatology presenting a history of multiple pigmented skin lesions that have been appearing over the last 30 years, with no prior treatment. Upon examination more than 50 hyperpigmented plaques and nodules on the face, left upper member and chest were identified, all compatible with pigmented BCC. Additional findings encountered were frontal bossing, chalazia of both superior eyelids, oligodontia and palmar pits. Imaging revealed falx cerebri calcification and bilateral radiolucid defects in the lower mandible. On further interrogation the patient stated that four brothers presented similar manifestations. With these findings and biopsy confirmation of suspected lesions the diagnosis of Gorlin syndrome was made. Treatment is ongoing, based on preventative education and a combination of physical treatment and surgical modalities to eradicate existing neoplasms.

Key Message: Gorlin Syndrome is a rare inherited skin disease that can lead to severe outcomes if not detected and treated as soon as possible. This case highlights this point as it exemplifies a rare case of very late diagnosis.