GENETICS AND GENODERMATOSES

CASE REPORT: WHOLE EXOME SEQUENCING HELPS IN ACCURATE MOLECULAR DIAGNOSIS IN SIBLINGS WITH A RARE CO-OCCURRENCE OF PATERNALLY INHERITED 22Q12DUPLICATION AND AUTOSOMAL RECESSIVE NON-SYNDROMIC ICHTHYOSIS

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Background: Autosomal recessive congenital ichthyosis (ARCI), a heterogeneous disorder of cornification of skin, encompasses three clinical subtypes: lamellar ichthyosis (LI; OMIM 242300); congenital ichthyosiform erythroderma (CIE; OMIM 242100); and harlequin ichthyosis (HI; OMIM 242500). LI has an incidence of approximately 1 in 250,000. Usually associated with mutations in the transglutaminase gene (TGM1), mutations in six other genes have, less frequently, been shown to be causative.

Observation: Two, 8 and 6-year-old siblings, born in a collodion membrane, presented with fish like scales all over the body. Karyotyping revealed duplication of the chromosome arm on 22q12+ in the father and two siblings. Both siblings were heat intolerant, photosensitive and hypohidrotic. Born uneventfully vaginally they were encased in a collodion membrane which was shed within a week of birth. There was no family history of any dermatoses.
Karyotyping of their parents and the siblings performed previously revealed duplication of the chromosome arm on 22q12+ in the father and two siblings. Whole exome sequencing revealed a homozygous p.Gly218Ser variation in TGM1; a variation reported earlier in an isolated Finnish population in association with autosomal recessive non-syndromic ichthyosis.

Key Message: This concurrence of a potentially benign 22q12+ duplication and LI, both rare individually, is reported here likely for the first time.