GENETICS AND GENODERMATOSES

# HOMOZYGOUS TGM1 MUTATION IN 6 UNRELATED PATIENTS WITH LAMELLAR ICHTHYOSIS FROM MALAYSIA

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Introduction : Autosomal recessive congenital ichthyosis (ARCI) is heterogeneous group of keratinization disorders . To date 9 causative genes have been identified.
Large part of ARCI is formed by lamellar icthyosis (LI) that mainly is caused by mutations in TGM1.
Mutations in TGM1 lead to decrease activity of TGase-1, with consequences such as defective cornified cell envelope and skin-barrier function.

Objective : To evaluate the genotype phenotype correlation of 6 adult patients with Lamellar Ichthyosis

Material and Method : Six adults patients with Lamellar Ichthyosis are identified from northern Malaysia . Mutation screening were performed for TGM1 gene by PCR amplification of each TGM1 exon and part of its flanking introns, followed by direct DNA sequencing.

Results and discussion : We detected homozygous mutation c.1166G>A (Arg389His) in the TGM1 gene in all 6 patients. To date, more than 135 TGM1 mutations, mostly missense and nonsense, identified to cause ARCI. The Arg389His has previously been reported in patients with LI from the US and Japan. The mutation is in the highly conserved residue located in the center of the catalytic core domain of TGase 1 peptide.
Conclusion : We report the identification of homozygous Arg389His mutation of TGM1 gene in six LI patients originated from Northern Malaysia. Further study is needed to establish the carrier status of the members of the patients' families, carrier and frequency in relevant community.