GENETICS AND GENODERMATOSES

GENETIC PRELIMINARY EXAMINATION: ADENOSINE TRIPHOSPHATE-BINDING CASSETTE A12 GENE EXON 34 AND 44 IN A CASE OF HARLEQUIN ICHTHYOSIS AND HER BOTH PARENTS

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Background: Harlequin ichthyosis (HI) is an extremely rare and the most severe form of autosomal recessive congenital ichthyosis. HI is caused by an adenosine triphosphate-binding cassette A12 (ABCA12) mutations which consist of 56 exons, with the most mutations found in exons 34 and 44, including in Malaysian races. No genetic analysis has been conducted on HI cases in Indonesian yet. The purpose of this case report is to initiate genetic HI testing of the Indonesian race, with emphasis on exon 34 and 44 analyzes which proven to be a “hot spot” on HI.

Observation: A 13-day-old girl neonate presented with thick, hyperkeratotic plate-like scales covering almost all parts of the body with deep fissures, prominent eclabium, bilateral ectropion, poorly formed and flattened ears and nose, hypoplasia of the fingers and feet at birth. A genetic examination was carried out from the patient and both parents due to mutation detection in exon 34 and 44. ABCA12 genetic examination in this case report was carried out on exons 34 and 44 based on the races of Malaysian and Singaporean. The results of ABCA12 gene examination in exon 34 and 44 from this patient and both parents were not obtained any mutation. At present, the patient is six months old and being regularly followed-up. Most of the hyperkeratotic scales have shed off and been replaced by neo-epithelial tissues.

Key message: Although no mutations are found, there is still a need for a whole-genome examination to be able to conclude the presence or absence of mutations in HI with Indonesian race, it might be the first complete examination case of HI.