GENETICS AND GENODERMATOSES

CORRELATION BETWEEN PHENOTYPE AND GENETIC MUTATION IN VARIOUS TYPE OF EPIDERMOLYSIS BULLOSA PATIENTS IN INDONESIA

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Introduction: Epidermolysis bullosa (EB), a group of genodermatosis with considerable clinical and genetic heterogeneity, has been divided into distinct subtypes depending on the level of tissue separation at the dermal-epidermal basement membrane zone. There are autosomal dominant and autosomal recessive forms. Clinically, EB presents with varying severity. Clinical diagnosis of EB subtypes is frequently inaccurate without genetic testing.

Objective: To assess the genetic mutations in our EB patients and further correlate with the clinical phenotype.

Materials and methods: We performed an analytic observational study which included all EB patients from January to December 2017. All patients were clinically examined to determine the type of EB. Two milliliter of blood was extracted from each patient for DNA isolation. Genotyping examination was done using Miseq Illumina system®, looking at all possible EB gene mutations.

Results: There were 10 EB patients included in this study. Genetic testing revealed mutations in COL7A1 gene in 8 patients, of which 6 were determined as dominant dystrophic EB (DDEB) and 2 patients were recessive dystrophic EB (RDEB). The last two patients showed mutations in KRT14 gene, confirming diagnosis of EB simplex (EBS). The initial clinical diagnosis of only 3 patients (30%) corresponded to the diagnosis obtained from genetic testing. Of the 7 patients whose clinical diagnosis did not correspond to genetic diagnosis, 3 patients with confirmed DDEB were previously diagnosed as EBS or junctional EB (JEB), 2 patients with confirmed RDEB were clinically diagnosed as DDEB or EBS, and the 2 patients with confirmed EBS were previously clinically diagnosed as JEB.

Conclusions: The use of genetic testing to confirm EB subtype is important as clinical assessments may be inaccurate, especially in younger patients who do not manifest all the clinical features of the EB subtype.