Complex Genetic Disorders, Genetic Susceptibility to Infections

AB071. Semaphorin 3D impact in Indonesian Hirschsprung patients

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Background: Hirschsprung disease (HSCR) is a heterogeneous genetic disorder characterized by absence of ganglion cells along the intestines, results in functional bowel obstruction in children. Recently, Semaphorin 3D (*SEMA3D*) gene has been implicated in the pathogenesis of intestinal ganglionosis. We aimed to conduct a mutation analysis of *SEMA3D* gene in HSCR patients in Indonesia, a genetically distinct group within Asia.

Methods: We ascertained 40 patients with HSCR of whom 27 and 13 were males and females, respectively. Subsequently, we performed direct sequencing to clarify the contribution of *SEMA3D* gene to HSCR development.

Results: All patients were sporadic HSCR with degree of aganglionosis as follows: short-segment in 39/40 (98%) patients and long-segment in 1/40 (2%) patients. Transanal endorectal pull-through (TEPT) has been the most common definitive surgery (54%), followed by Duhamel (21%), and Soave (14%). Mutation analysis of *SEMA3D* gene showed no rare variant, but one common variant in exon 17, rs7800072. The risk allele frequency at rs7800072 (C) among HSCR patients were 0.52.

Conclusions: This result implies that the *SEMA3D* gene may not have an effect in the molecular pathogenesis of HSCR, particularly in Indonesia. This study is the first report of *SEMA3D* gene in Asian ancestry. Further study with multicenter and a larger number of samples is necessary to clarify the results.

Keywords: Hirschsprung disease (HSCR); Semaphorin 3D (*SEMA3D*); Indonesia; Asian ancestry

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