RET single nucleotide polymorphism in Indonesians with sporadic HirschsprungÃƒÂ¢Ã¢Â‚Â¬Ã¢Â„Â¢s disease

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Abstract	The tyrosine kinase receptor RET, which is the protein product of the RET gene, is involved in the development of the mammalian nervous system that causes HirschsprungÃƒÂ¢Ã¢Â‚Â¬Ã¢Â„Â¢s disease (HSCR). RETs are cell surface molecules that are expressed in cells derived from the neural crest. The purpose of this study was to investigate the polymorphism of the RET gene in HSCR in the Yogyakarta population. Genomic DNA was extracted from surgically removed bowel tissues of 54 unrelated HSCR patients. Exon 2 of the RET gene was amplified by polymerase chain reaction (PCR) and analyzed by restriction fragment length polymorphism (RFLP). Molecular results were compared with clinical performance of Hirschsprung patients. RET polymorphism was detected in exon 2 in all of the 54 Indonesian HSCR patients. The allelic distribution of the c135GÃƒÂƒÃ‚Â A polymorphism in the RET exon 2 indicated that the A allele was more frequent in patients than in control individuals (chi-square test, p= 0.001). Thus the RET variant allele A is over-represented in patients affected with the HSCR phenotype. Polymorphism of exon 2 of the RET gene was found in sporadic HirschsprungÃƒÂ¢Ã¢Â‚Â¬Ã¢Â„Â¢s disease in the Yogyakarta population, which suggests that the RET gene plays important roles in the pathogenesis of HSCR.
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