

Table S1: Characteristics of the HSCR patients included in the study

Samples	Relationship	Gender	Aganglionosis	Associated anomalies/others	Ethnicity
HK9C* HK9A HK9B	Proband Father Mother	M M F	Long	Necrotizing enterocolitis; asthma	Chinese
HK96C* HK96A HK96B	Proband Father Mother	M M F	Total	Parathyroid nodules; bilateral hydrocele; necrotizing enterocolitis	Chinese
HK97C* HK97A HK97B	Proband Father Mother	M M F	Long	Congenital central hypoventilation syndrome (CCHS); necrotizing enterocolitis ; mild mental retardation; epilepsy	Chinese
HK164C HK164A HK164B	Proband Father Mother	M M F	Long	None; consanguineous parents	South-east Asian
HD09C* HD09A HD09B	Proband Father Mother	M M F	Long	None	Chinese
VH105C VH105A VH105B	Proband Father Mother	M M F	Long	None	Vietnamese
VH106C VH106A VH106B	Proband Father Mother	M M F	Long	None	Vietnamese
VH108C VH108A VH108B	Proband Father Mother	M M F	Long	None	Vietnamese
HK180C HK180A HK180B	Proband Father Mother	M M F	Long	None	Chinese

*Sample included in: MM Garcia-Barceló, CS Tang, *et al.* Genome-wide association study identifies *NRG1* as a susceptibility locus for Hirschsprung's disease. 2009. PNAS 106:2694-2699