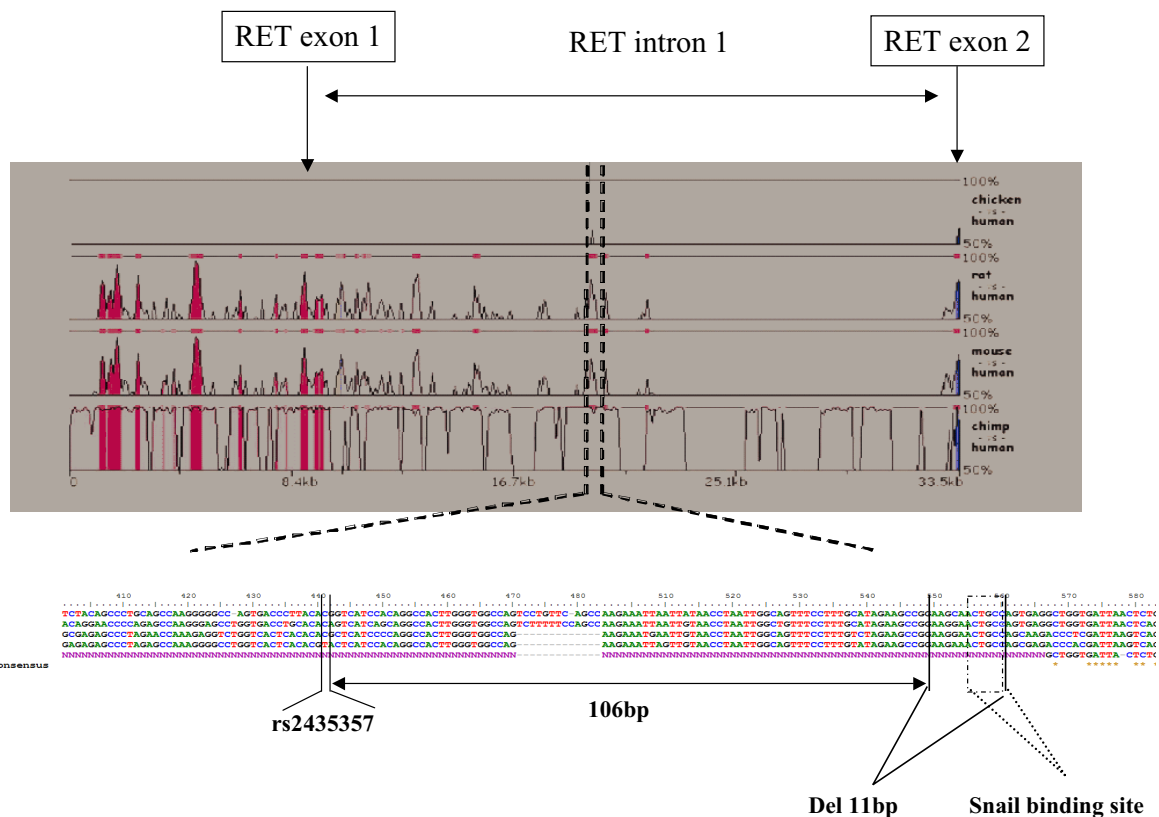


# Supporting Information

de Pontual et al. 10.1073/pnas.0901219106



**Fig. S1.** Considering the rs2435357 SNP lying in intron one of the *RET* gene, the T allele is the hypomorphic Hirschsprung disease (HSCR) predisposing *RET* allele, and the C is the WT type allele. del, the (AAGCACTGCC) 11-bp deletion identified in intron 1 of *RET* gene.



**Table S1. Ciliary proteins expressed in cultured human NCC**

Protein	Cellular localization
BBS1	Basal body/centrosome
BBS10	Basal body/centrosome
BBS2	Basal body/centrosome
BBS3	Basal body/centrosome
BBS4	Basal body/centrosome
BBS5	Basal body/centrosome
BBS6	Basal body/centrosome
BBS7	Basal body/centrosome
BBS8	Basal body/centrosome
BBS9	Basal body/centrosome
CCDC28B	Basal body/centrosome
CETN3	Basal body/centrosome
LRRC1	Basal body/centrosome
MKS1	Basal body/centrosome
MKS3	Basal body/centrosome
OFD1	Basal body/centrosome
TUBE1	Basal body/centrosome
TUBG1	Basal body/centrosome
TUBG2	Basal body/centrosome
PPP1CC	Central pair
SPAG6	Central pair
CALM3	Ciliary axoneme
CCDC146	Ciliary axoneme
Cys1	Ciliary axoneme
EFHC1	Ciliary axoneme
GLI2	Ciliary axoneme
HSPA1A	Ciliary axoneme
HYDIN	Ciliary axoneme
PPP2R1A	Ciliary axoneme
RIBC1	Ciliary axoneme
SSNA1	Ciliary axoneme
TEKT2	Ciliary axoneme
TEKT3	Ciliary axoneme
HTR1B	Ciliary membrane
HTR2C	Ciliary membrane
PDGFRA	Ciliary membrane
PKD1	Ciliary membrane
PKD2	Ciliary membrane
PKHD1	Ciliary membrane
SMO	Ciliary membrane
CROCC	Ciliary rootlet
GAS8	Dynein regulatory complex
ACTG1	Inner dynein arm
DNAH7	Inner dynein arm
DNALI1	Inner dynein arm
DYNLT1	Inner dynein arm
WDR63	Inner dynein arm
WDR78	Inner dynein arm
DNCH2	Intraflagellar transport
IFT122	Intraflagellar transport
IFT140	Intraflagellar transport
IFT172	Intraflagellar transport
IFT20	Intraflagellar transport
IFT52	Intraflagellar transport
IFT57	Intraflagellar transport
IFT72	Intraflagellar transport
IFT74	Intraflagellar transport
IFT80	Intraflagellar transport
IFT81	Intraflagellar transport
IFT88	Intraflagellar transport
KIF3A	Intraflagellar transport
KIF3B	Intraflagellar transport
KIFAP3	Intraflagellar transport
AK5	Outer dynein arm

Protein	Cellular localization
CCDC63	Outer dynein arm
DNAH5	Outer dynein arm
DNAH9	Outer dynein arm
DNAI1	Outer dynein arm
DNAI2	Outer dynein arm
DNAL1	Outer dynein arm
DYNLL2	Outer dynein arm
DYNLRB2	Outer dynein arm
TCTEX1D2	Outer dynein arm
DAAM1	PCP pathway
DVL1	PCP pathway
DVL2	PCP pathway
DVL3	PCP pathway
MAPK8	PCP pathway
ROCK1	PCP pathway
VANGL2	PCP pathway
RSHL3	Radial spoke
RSPH3	Radial spoke
TUBA1A	Tubulin
TUBB2C	Tubulin