Table S3: List of 117 ENS-related HSCR candidate genes

Pathways	Gene	Chromosome	Evidence	Reference
Others	*ALDH1A2*	15q22.1	Mouse (Absence EN)	[1]
Others	*ARHGEF3*	3p14.3	Expression	[2-3]
Ret signaling	*ARTN*	1p34.1	Mouse (Abnormal ENS morphology)	[4]
Notch signaling	*ASCL1*	12q23.2	Mouse (Absence EN)/Expression	[3,5-7]
Others	*CADM1*	11q23.2	Expression	[3,8]
Others	*CARTPT*	5q13.2	Expression	[3]
Others	*CBR1*	21q22.13	Expression	[9]
Others	*CDH2*	18q11.2	Expression	[3,10]
Semophorin signaling	*CRMP1*	4p16.1	Expression	[3,11]
Others	*CSTB*	21q22.3	Expression	[12]
Others	*CTNNAL1*	9q31.3	Expression	[3]
Others	*DCC*	18q21.2	Mouse (Absence submucosal ganglia)	[13]
Others	*DCX*	Xq22.3-q23	Expression	[3]
Notch signaling	*DLL1*	6q27	Not described	[6-7]
Notch signaling	*DLL3*	19q13.2	Not described	[6-7]
Others	*DLX1*	2q32	Expression	[3,14-15]
Others	*DPYSL3*	5q32	Expression	[3,16]
Others	*EBF3*	10q26.3	Expression	[3,17]
Others	*ECE1*	1p36	Human (Linkage)/Mouse (Absence EN)	[13, 18-19]
Ednrb signaling	*EDN3*	20q13	Human (Linkage)/Mouse (Absence EN)	[13,18,20-21]
Ednrb signaling	*EDNRB*	13q22	Human (Linkage/CNV)/Mouse (Absence EN)	[13,18,22-23]
Others	*ELAVL2*	9p21	Expression	[3,24]
Others	*ELAVL4*	1p34	Expression	[3,25]
Neuregulin signaling	*ERBB2*	17q12	Mouse (Abnormal ENS morphology)	[26-27]
Neuregulin signaling	*ERBB3*	12q13.2	Mouse (Abnormal ENS morphology)	[26-27]
Neuregulin signaling	*ERBB4*	2q33.3-q34	Human (CNV)	[28]
Others	*ETV1*	7p21.3	Expression	[3, 29]
Others	*FGF13*	Xq26.3	Expression	[3,30]
Others	*GAP43*	3q13.1-q13.2	Expression	[3,31]
Ret signaling	*GDNF*	5p13	Human (Linkage)/Mouse (Absence EN)/Expression	[13,18,32-35]
Ret signaling	*GFRA1*	10q25	Human (1 patient)/Mouse (Absence EN)/Expression	[3-4, 34,36]
Ret signaling	*GFRA2*	8p21.3	Mouse (Abnormal ENS morphology)	[4]
Ret signaling	*GFRA3*	5q11.2	Mouse (Abnormal sympathetic system)	[4]
Ret signaling	*GFRA4*	20p13	Not described	[4]
Hedgehog signaling	*GLI1*	12q13.3	Mouse (Abnormal intestinal morphology)	[37-38]
Hedgehog signaling	*GLI2*	2q14	Mouse (Abnormal intestinal morphology)	[37-38]

Table S3: List of 117 ENS-related HSCR candidate genes

Pathways	Gene	Chromosome	Evidence	Reference
Hedgehog signaling	*GLI3*	7p14	Mouse (Abnormal intestinal morphology)	[37-38]
Others	*GNG2*	14q21	Expression	[3]
Others	*GNG3*	11p11	Expression	[3,39]
Others	*GRB10*	7p12.2	Human (Linkage)	[34]
Hedgehog signaling	*HES1*	3q29	Mouse (Abnormal intestinal morphology)	[37-38]
Others	*HLX*	1q41	Mouse (Hypoaganglionosis)	[13]
Others	*HMP19*	5q35.2	Expression	[3]
Others	*HMX3*	10q26.13	Expression	[3,40]
Others	*HOXB5*	17q21.3	Expression	[3,41]
Others	*HOXD4*	2q31.1	Expression	[3,42]
Others	*IFNGR2*	21q22.11	Expression	[43]
Hedgehog signaling	*IHH*	2q35	Mouse (Absence EN)	[37-38]
Others	*IKBKAP*	9q31.3	Human (Co-Expression)	[44]
Others	*IL10RB*	21q22.11	Expression	[45]
Others	*ITGB1*	10p11.22	Mouse (Absence EN)	[13]
Notch signaling	*JAG1*	20p12.1	Not described	[6-7]
Notch signaling	*JAG2*	14q32.33	Not described	[6-7]
Others	*KIAA1279*	10q21	Human (Linkage/GSM syndrome)	[46]
Hedgehog signaling	*KLF4*	9q31	Mouse (Abnormal intestinal morphology)	[37-38]
Others	*L1CAM*	Xq28	Human (Hydrocephalus)/Mouse (Delayed NCC differentiation)/Expression	[3,47-49]
Others	*MAB21L1*	13q13	Expression	[3,50]
Others	*MAPK10*	4q22.1-q23	Expression	[3,51]
Others	*MAPT*	17q21.1	Expression	[3]
Others	*MLLT11*	1q21	Expression	[3,52]
Others	*NAV2*	11p15.1	Human (Exome)	[53]
Others	*NKX2-1*	14q13	Human (Case report)	[13,18]
Notch signaling	*NOTCH1*	9q34	Not described	[6-7]
Notch signaling	*NOTCH2*	6q27	Not described	[6-7]
Notch signaling	*NOTCH3*	19p13.12	Not described	[6-7]
Neuregulin signaling	*NRG1*	8p12	Human (GWAS)/Mouse (Abnormal NCC migration)	[54]
Neuregulin signaling	*NRG3*	10q22-q23	Human (CNV/Exome)	[28,55]
Semophorin signaling	*NRP1*	10p11.22	Not described	[56-57]
Others	*NRTN*	19p13	Human (Linkage)/Mouse (Abnormal ENS)	[58]
Others	*NTF3*	1p36.11	Mouse (Reduced enteric ganglia)	[13]
Others	*NTRK3*	15q25	Mouse (Reduced enteric ganglia)	[13]
Ret signaling	*PAX3*	2q36	Human (Exome)/Mouse (Absence EN)	[4,53]

Table S3: List of 117 ENS-related HSCR candidate genes

Pathways	Gene	Chromosome	Evidence	Reference
Others	*PCDHA1*	5q31	Expression	[3,59]
Others	*PFKL*	21q22.3	Expression	[60]
Others	*PHACTR4*	1p35.3	Mouse	[61-62]
Others	*PHOX2A*	11q13.2	Expression	[3,63]
Others	*PHOX2B*	4p13	Human (Haddad syndrome)/Mouse (Abnormal ENS)/Expression	[3,64-65]
Semophorin signaling	*PLXNA1*	3q21.3	Not described	[56-57]
Semophorin signaling	*PLXNB1*	3p21	Human (Linkage)	[66-67]
Notch signaling	*POFUT1*	20q11.21	Mouse (Absence EN)	[6-7]
Prokinecticin signaling	*PROK1*	1p13	Not described	[68-69]
Prokinecticin signaling	*PROK2*	3p13	Not described	[68-69]
Prokinecticin signaling	*PROKR1*	2p14	Not described	[68-69]
Prokinecticin signaling	*PROKR2*	20p12	Not described	[68-69]
Others	*PRPH*	12q12-q13	Expression	[3,70]
Ret signaling	*PSPN*	19p13.3	Not described	[4]
Hedgehog signaling	*PTCH1*	9q22.32	Mouse (Abnormal intestinal morphology)	[37-38]
Ret signaling	*RET*	10q11	Human (Linkage/CNV/Exome)/Mouse (Absence EN)	[3,53,71-72]
Others	*SALL4*	20q13.2	Mouse (Absence EN)	[13]
Others	*SCG3*	15q21	Expression	[3]
Semophorin signaling	*SEMA3A*	7p12	Human (Association)/Mouse (Abnormal ENS morphology)	[53,73]
Semophorin signaling	*SEMA3C*	7p12	Human (GWAS)/Others (Not described)	[56-57]
Semophorin signaling	*SEMA3D*	7p12	Human (GWAS)/Others (Not described)	[56-57]
Others	*SERPINI1*	3q26.1	Expression	[3,74-75]
Hedgehog signaling	*SHH*	7q36	Mouse (Ectopic enteric ganglia formation)	[37-38]
Hedgehog signaling	*SMO*	7q32	Mouse (Abnormal neural crest cell migration)	[37-38]
Others	*SOD1*	21q22.11	Expression	[76-77]
Others	*SON*	21q22.11	Expression	[78]
Others	*SOX10*	22q13	Human (Linkage/WS4)/Mouse (Absence EN)	[3,79]
Others	*SOX2*	3q26.3-q27	Expression	[3,80]
Others	*SPRY2*	13q31.1	Mouse (Increased EN)	[13]
Others	*STMN2*	8q21.13	Expression	[3,81]
Others	*STMN3*	20q13.3	Expression	[3]
Hedgehog signaling	*SUFU*	10q24.32	Mouse (Abnormal neural tube morphology)	[37-38]
Others	*SYT11*	1q21.2	Expression	[3,82]
Others	*TAGLN3*	3q13.2	Expression	[3]
Others	*TBX3*	12q24.1	Expression	[83-84]
Others	*TFF3*	21q22.3	Expression	[3,85-86]

Table S3: List of 117 ENS-related HSCR candidate genes

Pathways	Gene	Chromosome	Evidence	Reference
Others	*TGFB2*	1q41	Expression	[3,87]
Others	*TMEFF2*	2q32.3	Expression	[3,88]
Others	*TREX1*	3p21.31	Human (Linkage)	[66-67]
Others	*TTC3*	21q22.2	Expression	[89]
Others	*TUBB3*	16q24.3	Expression	[3,90]
Others	*UCHL1*	4p14	Expression	[3,91]
Others	*VIP*	6q25	Expression	[3,92]
Others	*ZEB2*	2q22	Human (Linkage/MW syndrome/CNV)/Mouse (Abnormal NCC migration)	[93-95]
Others	*ZIC2*	13q32	Mouse	[96-97]

Notes: updated gene symbols used for ZFHX1B (replaced by ZEB2), TRKC (replaced by NTRK3) and RALDH2 (replaced by ALDH1A2); 'EN' means enteric neurons; 'NCC' means neural crest cells

References to TableS3

	Authors	***Title***	***Journal***
1	Anderson, R. B., D. F. Newgreen, et al. (2006).	Neural crest and the development of the enteric nervous system.	Adv Exp Med Biol 589: 181-196.
2	Thiesen, S., S. Kubart, et al. (2000).	Isolation of two novel human RhoGEFs, ARHGEF3 and ARHGEF4, in 3p13-21 and 2q22.	Biochem Biophys Res Commun 273(1): 364-369.
3	Heanue, T. A. and V. Pachnis (2006).	Expression profiling the developing mammalian enteric nervous system identifies marker and candidate Hirschsprung disease genes.	Proc Natl Acad Sci U S A 103(18): 6919-6924.
4	Pachnis, V., P. Durbec, et al. (1998).	Role Of the RET signal transduction pathway in development of the mammalian enteric nervous system.	Am J Physiol 275(2 Pt 1): G183-186.
5	Huang, H. S., G. M. Kubish, et al. (2010).	Direct transcriptional induction of Gadd45gamma by Ascl1 during neuronal differentiation.	Mol Cell Neurosci 44(3): 282-296.
6	Okamura, Y. and Y. Saga (2008).	Notch signaling is required for the maintenance of enteric neural crest progenitors.	Development 135(21): 3555-3565.
7	Morrison, S. J., S. E. Perez, et al. (2000).	Transient Notch activation initiates an irreversible switch from neurogenesis to gliogenesis by neural crest stem cells.	Cell 101(5): 499-510.
8	Hagiyama, M., N. Ichiyanagi, et al. (2009).	Expression of a soluble isoform of cell adhesion molecule 1 in the brain and its involvement in directional neurite outgrowth.	Am J Pathol 174(6): 2278-2289.
9	Sultan, M., I. Piccini, et al. (2007).	Gene expression variation in Down's syndrome mice allows prioritization of candidate genes.	Genome Biol 8(5): R91.
10	Hermiston, M. L. and J. I. Gordon (1995).	Inflammatory bowel disease and adenomas in mice expressing a dominant negative N-cadherin.	Science 270(5239): 1203-1207.

11	Schmidt, E. F., S. O. Shim, et al. (2008).	Release of MICAL autoinhibition by semaphorin-plexin signaling promotes interaction with collapsin response mediator protein.	J Neurosci 28(9): 2287-2297.
12	Brannvall, K., H. Hjelm, et al. (2003).	Cystatin-B is expressed by neural stem cells and by differentiated neurons and astrocytes.	Biochem Biophys Res Commun 308(2): 369-374.
13	Heanue, T. A. and V. Pachnis (2007).	Enteric nervous system development and Hirschsprung's disease: advances in genetic and stem cell studies.	Nat Rev Neurosci 8(6): 466-479.
14	Simeone, A., D. Acampora, et al. (1994).	Cloning and characterization of two members of the vertebrate Dlx gene family.	Proc Natl Acad Sci U S A 91(6): 2250-2254.
15	Cobos, I., U. Borello, et al. (2007).	Dlx transcription factors promote migration through repression of axon and dendrite growth.	Neuron 54(6): 873-888.
16	Hamajima, N., K. Matsuda, et al. (1996).	A novel gene family defined by human dihydropyrimidinase and three related proteins with differential tissue distribution.	Gene 180(1-2): 157-163.
17	Zardo, G., M. I. Tiirikainen, et al. (2002).	Integrated genomic and epigenomic analyses pinpoint biallelic gene inactivation in tumors.	Nat Genet 32(3): 453-458.
18	Amiel, J. and S. Lyonnet (2001).	Hirschsprung disease, associated syndromes, and genetics: a review.	J Med Genet 38(11): 729-739.
19	Hofstra, R. M., O. Valdenaire, et al. (1999).	A loss-of-function mutation in the endothelin-converting enzyme 1 (ECE-1) associated with Hirschsprung disease, cardiac defects, and autonomic dysfunction.	Am J Hum Genet 64(1): 304-308.
20	Bidaud, C., R. Salomon, et al. (1997).	Mutations of the endothelin-3 gene in isolated and syndromic forms of Hirschsprung disease.	Gastroenterol Clin Biol 21(8-9): 548-554.
21	Sanchez-Mejias, A., R. M. Fernandez, et al. (2009).	Contribution of RET, NTRK3 and EDN3 to the expression of Hirschsprung disease in a multiplex family.	J Med Genet 46(12): 862-864.
22	Carrasquillo, M. M., A. S. McCallion, et al. (2002).	Genome-wide association study and mouse model identify interaction between RET and EDNRB pathways in Hirschsprung disease.	Nat Genet 32(2): 237-244.
23	Kenny, S. E., R. M. Hofstra, et al. (2000).	Reduced endothelin-3 expression in sporadic Hirschsprung disease.	Br J Surg 87(5): 580-585.
24	Han, J., J. F. Knops, et al. (1996).	Localization of human elav-like neuronal protein 1 (Hel-N1) on chromosome 9p21 by chromosome microdissection polymerase chain reaction and fluorescence in situ hybridization.	Genomics 36(1): 189-191.
25	Akamatsu, W., H. Fujihara, et al. (2005).	The RNA-binding protein HuD regulates neuronal cell identity and maturation.	Proc Natl Acad Sci U S A 102(12): 4625-4630.
26	Paratore, C., D. E. Goerich, et al. (2001).	Survival and glial fate acquisition of neural crest cells are regulated by an interplay between the transcription factor Sox10 and extrinsic combinatorial signaling.	Development 128(20): 3949-3961.
27	Britsch, S. (2007).	The neuregulin-I/ErbB signaling system in development and disease.	Adv Anat Embryol Cell Biol 190: 1-65.
28	Garcia-Barcelo, M. M., C. S. Tang, et al. (2009).	Genome-wide association study identifies NRG1 as a susceptibility locus for Hirschsprung's disease.	Proc Natl Acad Sci U S A 106(8): 2694-2699.
29	Flames, N. and O. Hobert (2009).	Gene regulatory logic of dopamine neuron differentiation.	Nature 458(7240): 885-889.

30	Smallwood, P. M., I. Munoz-Sanjuan, et al. (1996).	Fibroblast growth factor (FGF) homologous factors: new members of the FGF family implicated in nervous system development.	Proc Natl Acad Sci U S A 93(18): 9850-9857.
31	Strittmatter, S. M., C. Fankhauser, et al. (1995).	Neuronal pathfinding is abnormal in mice lacking the neuronal growth cone protein GAP-43.	Cell 80(3): 445-452.
32	Salomon, R., T. Attie, et al. (1996).	Germline mutations of the RET ligand GDNF are not sufficient to cause Hirschsprung disease.	Nat Genet 14(3): 345-347.
33	Ivanchuk, S. M., S. M. Myers, et al. (1996).	De novo mutation of GDNF, ligand for the RET/GDNFR-alpha receptor complex, in Hirschsprung disease.	Hum Mol Genet 5(12): 2023-2026.
34	Angrist, M., S. Bolk, et al. (1998).	Genomic structure of the gene for the SH2 and pleckstrin homology domain-containing protein GRB10 and evaluation of its role in Hirschsprung disease.	Oncogene 17(23): 3065-3070.
35	Fernandez, R. M., M. Ruiz-Ferrer, et al. (2008).	Polymorphisms in the genes encoding the 4 RET ligands, GDNF, NTN, ARTN, PSPN, and susceptibility to Hirschsprung disease.	J Pediatr Surg 43(11): 2042-2047.
36	Myers, S. M., R. Salomon, et al. (1999).	Investigation of germline GFR alpha-1 mutations in Hirschsprung disease.	J Med Genet 36(3): 217-220.
37	Fu, M., V. C. Lui, et al. (2004).	Sonic hedgehog regulates the proliferation, differentiation, and migration of enteric neural crest cells in gut.	J Cell Biol 166(5): 673-684.
38	Reichenbach, B., J. M. Delalande, et al. (2008).	Endoderm-derived Sonic hedgehog and mesoderm Hand2 expression are required for enteric nervous system development in zebrafish.	Dev Biol 318(1): 52-64.
39	Hurowitz, E. H., J. M. Melnyk, et al. (2000).	Genomic characterization of the human heterotrimeric G protein alpha, beta, and gamma subunit genes.	DNA Res 7(2): 111-120.
40	Bober, E., C. Baum, et al. (1994).	A novel NK-related mouse homeobox gene: expression in central and peripheral nervous structures during embryonic development.	Dev Biol 162(1): 288-303.
41	Fu, M., V. C. Lui, et al. (2003).	HOXB5 expression is spatially and temporarily regulated in human embryonic gut during neural crest cell colonization and differentiation of enteric neuroblasts.	Dev Dyn 228(1): 1-10.
42	Mavilio, F., A. Simeone, et al. (1986).	Differential and stage-related expression in embryonic tissues of a new human homoeobox gene.	Nature 324(6098): 664-668.
43	Murai, M., O. Turovskaya, et al. (2009).	Interleukin 10 acts on regulatory T cells to maintain expression of the transcription factor Foxp3 and suppressive function in mice with colitis.	Nat Immunol 10(11): 1178-1184.
44	Bolk, S., A. Pelet, et al. (2000).	A human model for multigenic inheritance: phenotypic expression in Hirschsprung disease requires both the RET gene and a new 9q31 locus.	Proc Natl Acad Sci U S A 97(1): 268-273.
45	Glocker, E. O., D. Kotlarz, et al. (2009).	Inflammatory bowel disease and mutations affecting the interleukin-10 receptor.	N Engl J Med 361(21): 2033-2045.
46	Brooks, A. S., A. M. Bertoli-Avella, et al. (2005).	Homozygous nonsense mutations in KIAA1279 are associated with malformations of the central and enteric nervous systems.	Am J Hum Genet 77(1): 120-126.
47	Griseri, P., Y. Vos, et al. (2009).	Complex pathogenesis of Hirschsprung's disease in a patient with hydrocephalus, vesico-ureteral reflux and a balanced translocation t(2;17)(q13;q11).	Eur J Hum Genet 17(4): 483-490.
48	Turner, K. N., M. Schachner, et al. (2009).	Cell adhesion molecule L1 affects the rate of differentiation of enteric neurons in the developing gut.	Dev Dyn 238(3): 708-715.

49	Kenwrick, S., A. Watkins, et al. (2000).	Neural cell recognition molecule L1: relating biological complexity to human disease mutations.	Hum Mol Genet 9(6): 879-886.
50	Mariani, M., D. Baldessari, et al. (1999).	Two murine and human homologs of mab-21, a cell fate determination gene involved in Caenorhabditis elegans neural development.	Hum Mol Genet 8(13): 2397-2406.
51	Gupta, S., T. Barrett, et al. (1996).	Selective interaction of JNK protein kinase isoforms with transcription factors.	EMBO J 15(11): 2760-2770.
52	Tse, W., W. Zhu, et al. (1995).	A novel gene, AF1q, fused to MLL in t(1;11) (q21;q23), is specifically expressed in leukemic and immature hematopoietic cells.	Blood 85(3): 650-656.
53	T. Turner, K. D. Nguyen, et al. (2013).	The burden of coding, non-coding and chromosomal mutations in Hirschsprung disease	ASHG 2013 abstract; http://www.ashg.org/2013meeting/abstracts/fulltext/f1301217...
54	Garcia-Barcelo, M. M., C. S. Tang, et al. (2009).	Genome-wide association study identifies NRG1 as a susceptibility locus for Hirschsprung's disease.	Proc Natl Acad Sci U S A 106(8): 2694-2699.
55	Yang J, Duan S, et al. (2013)	Exome sequencing identified NRG3 as a novel susceptible gene of Hirschsprung's disease in a Chinese population.	Mol Neurobiol 47(3): 957-966
56	Jiang, Q., S. Arnold etal (2015)	Functional Loss of Semaphorin 3C and/or Semaphorin 3D and their Epistatic Interaction with Ret are Critical to Hirschsprung Disease Liability	Am J Hum Genet 96: 581-596.
57	Kruger, R. P., J. Aurandt, et al. (2005).	Semaphorins command cells to move.	Nat Rev Mol Cell Biol 6(10): 789-800.
58	Doray, B., R. Salomon, et al. (1998).	Mutation of the RET ligand, neurturin, supports multigenic inheritance in Hirschsprung disease.	Hum Mol Genet 7(9): 1449-1452.
59	Sugino, H., S. Hamada, et al. (2000).	Genomic organization of the family of CNR cadherin genes in mice and humans.	Genomics 63(1): 75-87.
60	Elson, A., D. Levanon, et al. (1994).	Overexpression of liver-type phosphofructokinase (PFKL) in transgenic-PFKL mice: implication for gene dosage in trisomy 21.	Biochem J 299 (Pt 2): 409-415.
61	Allen, P. B., A. T. Greenfield, et al. (2004).	Phactrs 1-4: A family of protein phosphatase 1 and actin regulatory proteins.	Proc Natl Acad Sci U S A 101(18): 7187-7192.
62	Kim, T. H., J. Goodman, et al. (2007).	Phactr4 regulates neural tube and optic fissure closure by controlling PP1-, Rb-, and E2F1-regulated cell-cycle progression.	Dev Cell 13(1): 87-102.
63	Sasaki, A., M. Kanai, et al. (2003).	Molecular analysis of congenital central hypoventilation syndrome.	Hum Genet 114(1): 22-26.
64	Garcia-Barcelo, M., M. H. Sham, et al. (2003).	Association study of PHOX2B as a candidate gene for Hirschsprung's disease.	Gut 52(4): 563-567.
65	Ou-Yang, M. C., S. N. Yang, et al. (2007).	Concomitant existence of total bowel aganglionosis and congenital central hypoventilation syndrome in a neonate with PHOX2B gene mutation.	J Pediatr Surg 42(2): e9-11.
66	Gabriel, S. B., R. Salomon, et al. (2002).	Segregation at three loci explains familial and population risk in Hirschsprung disease.	Nat Genet 31(1): 89-93.
67	Garcia-Barcelo, M. M., P. Y. Fong, et al. (2008).	Mapping of a Hirschsprung's disease locus in 3p21.	Eur J Hum Genet 16(7): 833-840.

68	Ngan, E. S., C. K. Shum, et al. (2008).	Prokineticin-1 (Prok-1) works coordinately with glial cell line-derived neurotrophic factor (GDNF) to mediate proliferation and differentiation of enteric neural crest cells.	Biochim Biophys Acta 1783(3): 467-478.
69	Ngan, E. S., K. Y. Lee, et al. (2007).	Prokineticin-1 modulates proliferation and differentiation of enteric neural crest cells.	Biochim Biophys Acta 1773(4): 536-545.
70	Oblinger, M. M., J. Wong, et al. (1989).	Axotomy-induced changes in the expression of a type III neuronal intermediate filament gene.	J Neurosci 9(11): 3766-3775.
71	Edery, P., S. Lyonnet, et al. (1994).	Mutations of the RET proto-oncogene in Hirschsprung's disease.	Nature 367(6461): 378-380.
72	Emison, E. S., A. S. McCallion, et al. (2005).	A common sex-dependent mutation in a RET enhancer underlies Hirschsprung disease risk.	Nature 434(7035): 857-863.
73	Behar, O., J. A. Golden, et al. (1996).	Semaphorin III is needed for normal patterning and growth of nerves, bones and heart.	Nature 383(6600): 525-528.
74	Stoeckli, E. T., P. F. Lemkin, et al. (1989).	Identification of proteins secreted from axons of embryonic dorsal-root-ganglia neurons.	Eur J Biochem 180(2): 249-258.
75	Lee, M. S., D. H. Jun, et al. (2006).	Selection of neural differentiation-specific genes by comparing profiles of random differentiation.	Stem Cells 24(8): 1946-1955.
76	Moldrich, R. X., L. Dauphinot, et al. (2009).	Proliferation deficits and gene expression dysregulation in Down's syndrome (Ts1Cje) neural progenitor cells cultured from neurospheres.	J Neurosci Res 87(14): 3143-3152.
77	Le Pecheur, M., E. Bourdon, et al. (2005).	Oxidized SOD1 alters proteasome activities in vitro and in the cortex of SOD1 overexpressing mice.	FEBS Lett 579(17): 3613-3618.
78	Wynn, S. L., R. A. Fisher, et al. (2000).	Organization and conservation of the GART/SON/DONSON locus in mouse and human genomes.	Genomics 68(1): 57-62.
79	Southard-Smith, E. M., L. Kos, et al. (1998).	Sox10 mutation disrupts neural crest development in Dom Hirschsprung mouse model.	Nat Genet 18(1): 60-64.
80	Bylund, M., E. Andersson, et al. (2003).	Vertebrate neurogenesis is counteracted by Sox1-3 activity.	Nat Neurosci 6(11): 1162-1168.
81	Bahn, S., M. Mimmack, et al. (2002).	Neuronal target genes of the neuron-restrictive silencer factor in neurospheres derived from fetuses with Down's syndrome: a gene expression study.	Lancet 359(9303): 310-315.
82	Craxton, M. (2001).	Genomic analysis of synaptotagmin genes.	Genomics 77(1-2): 43-49.
83	Han, J., P. Yuan, et al. (2010).	Tbx3 improves the germ-line competency of induced pluripotent stem cells.	Nature 463(7284): 1096-1100.
84	Paulsen, F. P., C. W. Woon, et al. (2008).	Intestinal trefoil factor/TFF3 promotes re-epithelialization of corneal wounds.	J Biol Chem 283(19): 13418-13427.
85	Mashimo, H., D. C. Wu, et al. (1996).	Impaired defense of intestinal mucosa in mice lacking intestinal trefoil factor.	Science 274(5285): 262-265.
86	Bottner, M., K. Krieglstein, et al. (2000).	The transforming growth factor-betas: structure, signaling, and roles in nervous system development and functions.	J Neurochem 75(6): 2227-2240.

87	Krieglstein, K., J. Strelau, et al. (2002).	TGF-beta and the regulation of neuron survival and death.	J Physiol Paris 96(1-2): 25-30.
88	Eib, D. W., T. M. Holling, et al. (2000).	Expression of the follistatin/EGF-containing transmembrane protein M7365 (tomoregulin-1) during mouse development.	Mech Dev 97(1-2): 167-171.
89	Tsukahara, F., M. Hattori, et al. (1996).	Identification and cloning of a novel cDNA belonging to tetratricopeptide repeat gene family from Down syndrome-critical region 21q22.2.	J Biochem 120(4): 820-827.
90	Mongroo, P. S. and A. K. Rustgi (2010).	The role of the miR-200 family in epithelial-mesenchymal transition.	Cancer Biol Ther 10(3): 219-222.
91	Sakurai, M., K. Ayukawa, et al. (2006).	Ubiquitin C-terminal hydrolase L1 regulates the morphology of neural progenitor cells and modulates their differentiation.	J Cell Sci 119(Pt 1): 162-171.
92	Larsson, L. T. (1994).	Hirschsprung's disease--immunohistochemical findings.	Histol Histopathol 9(3): 615-629.
93	Wakamatsu, N., Y. Yamada, et al. (2001).	Mutations in SIP1, encoding Smad interacting protein-1, cause a form of Hirschsprung disease.	Nat Genet 27(4): 369-370.
94	Amiel, J., Y. Espinosa-Parrilla, et al. (2001).	Large-scale deletions and SMADIP1 truncating mutations in syndromic Hirschsprung disease with involvement of midline structures.	Am J Hum Genet 69(6): 1370-1377.
95	Van de Putte, T., A. Francis, et al. (2007).	Neural crest-specific removal of Zfhx1b in mouse leads to a wide range of neurocristopathies reminiscent of Mowat-Wilson syndrome.	Hum Mol Genet 16(12): 1423-1436.
96	Nagai, T., J. Aruga, et al. (1997).	The expression of the mouse Zic1, Zic2, and Zic3 gene suggests an essential role for Zic genes in body pattern formation.	Dev Biol 182(2): 299-313.
97	Sanchez-Camacho, C. and P. Bovolenta (2008).	Autonomous and non-autonomous Shh signalling mediate the in vivo growth and guidance of mouse retinal ganglion cell axons.	Development 135(21): 3531-3541.