Supplementary File 1

TADs structure and characteristics of associated genes in probands with phenotype not explained by the directly disrupted gene(s)

Figure: Visual representation of TADs. The colours next to the genes, represent DOMINO score and pLI score, respectively. On the upper part of each figure the colours represent the number of interactions between loci in the genome

Table: Topologically Associating Domain (TADs) structure in the proximity of BCT breakpoints

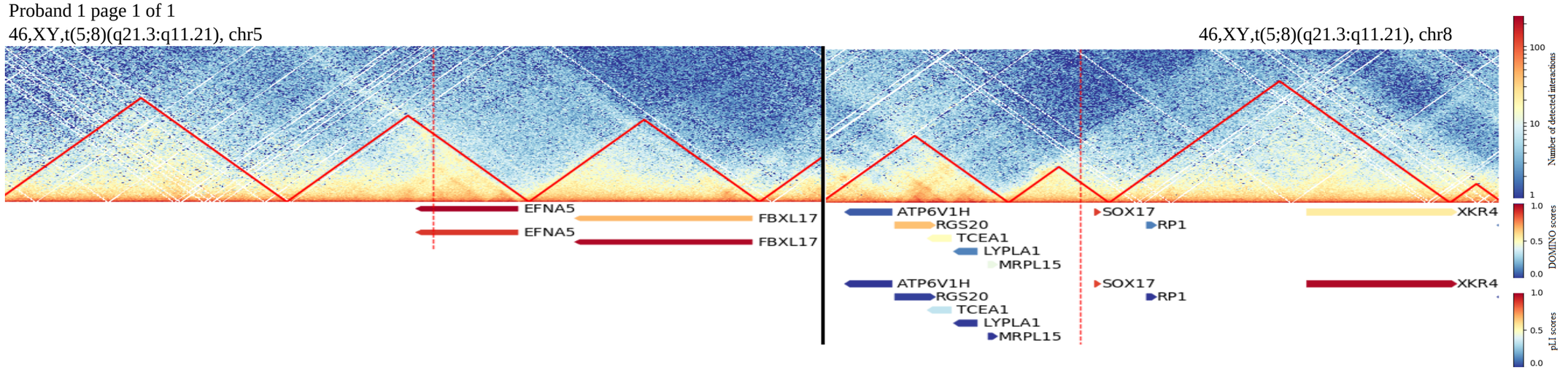
Domain – 0: same as breakpoint, +1: adjacent to breakpoint towards the telomere, -1 : adjacent to breakpoint towards the centromere.
Distance from breakpoint- distance vs. 5' or 3' end of the coding sequence of the gene (whatever shorter).

OMIM#, disease, inheritance -Information about inheritance from OMIM (https://omim.org/); AD - Autosomal dominant inheritance, AR- Autosomal recessive inheritance.

pLI - from http://exac.broadinstitute.org/
DOMINO – The score taken from DOMINO database (Quinodoz M, Royer-Bertrand B, Cisarova K, Di Gioia SA, Superti-Furga A, Rivolta C. DOMINO: Using Machine Learning to Predict Genes Associated with Dominant Disorders. Am J Hum Genet 2017;101(4):623-29. doi: 10.1016/j.ajhg.2017.09.001)
Mouse phenotype - from http://www.informatics.jax.org

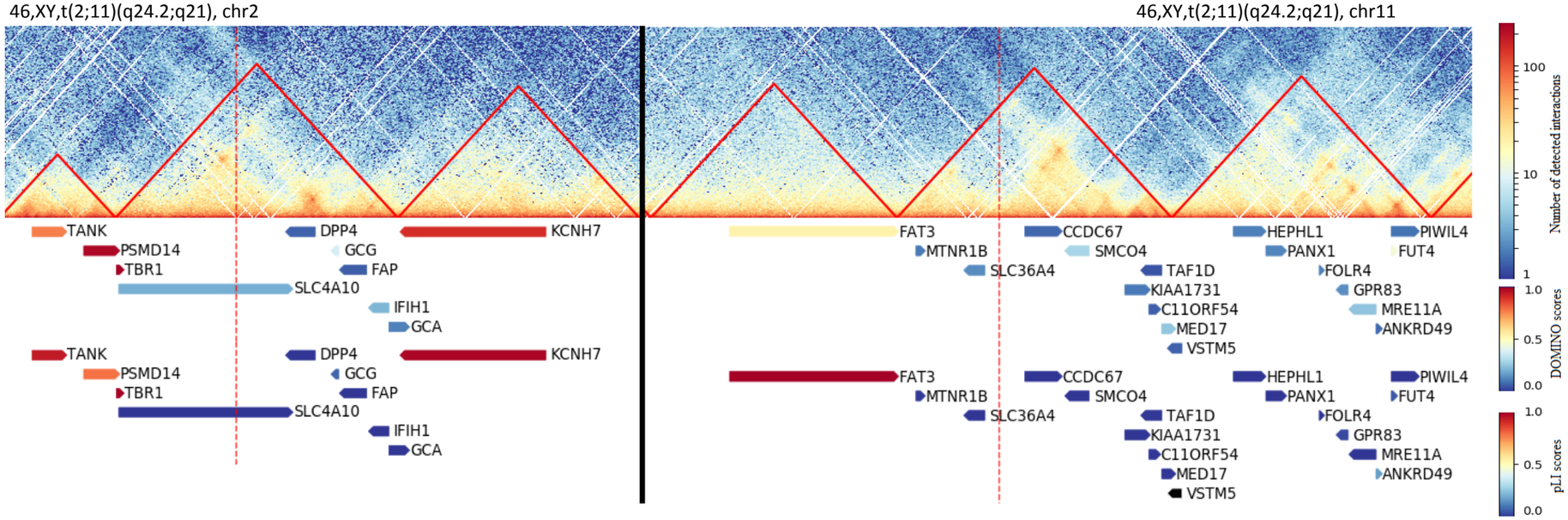
46,XY,t(5;8)(q21.3:q11.21), chr5

46,XY,t(5;8)(q21.3:q11.21), chr8



Breakpoint	**Gene**	**Distance from breakpoint (bp)/domain**	**OMIM#, disease, inheritance**	**pLI**	**DOMINO**	**Comments** (HGMD, mouse phenotype)
46,XY,t(5;8)(q21.3:q11.21), chr5	*FBXL17* (MIM: 609083), F-box and leucine-rich repeat protein 17	+446,390 / +1	NA	0,98	0,68	low confidence, early onset Parkinson [1]
46,XY,t(5;8)(q21.3:q11.21), chr8	*SOX17* (MIM: 610928), SRY-related hmg-box gene 17	+43,736/ 0	Vesicoureteral reflux (MIM: 613674), AD	0,87	0,88	NA
	RP1 (MIM: 603937), Retinitis pigmentosa 1	+201,868/+1	Retinitis pigmentosa (MIM: 180100), AD & AR	0	0,12	NA
	XKR4 (NA), XK Related 4	+688,190/+1	NA	0,98	0,56	NA
	MRPL15 (MIM: 611828), Mitochondrial ribosomal protein	-166,381/-1	NA	0	0,43	NA
	LYPLA1 (MIM: 605599), Lysophospholipase I	-312,265/-1	NA	0,01	0,13	NA
	TCEA1 (MIM: 601425), Transcription elongation factor A1	-391,753/-1	NA	0,34	0,5	Mouse: liver hypoplasia, defective erythropoesis, embryonic mortality
	RGS20 (MIM: 607193), Regulator of G protein signaling	-454,979/-1	NA	0,02	0,66	NA
	ATP6V1H (MIM: 608861), ATPase, H+ transporting	-570,724/-1	NA	0	0,06	Mouse: Decreased bone formation, embryonic mortality

1.Kun-Rodrigues C, Ganos C, Guerreiro R, Schneider SA, Schulte C, Lesage S, Darwent L, Holmans P, Singleton A, International Parkinson's Disease Genomics C, Bhatia K, Bras J. A systematic screening to identify de novo mutations causing sporadic early-onset Parkinson's disease. Hum Mol Genet 2015;24(23):6711-20. doi: 10.1093/hmg/ddv376



Breakpoint	**Gene**	**Distance from breakpoint (bp)/domain**	**OMIM#, disease, inheritance**	**pLI**	**DOMINO**	**Comments** (HGMD, mouse phenotype)
46,XY,t(2;11)(q24.2;q21), chr2	*DPP4* (MIM: 102720), Dipeptidyl peptidase IV	+181,050/0	NA	0	0,01	Autism spectrum disorder, autosomal
	GCG (MIM: 138030), Glucagon	+331,691/0	NA	0,08	0,38	Mouse: Hyperproglucagonemia
	FAP (MIM: 600403), Fibroblast activation	+359,493/0	NA	0	0,06	Loss of enzyme activity[3]
	IFIH1 (MIM: 606951), Interferon-induced	+455,888/0	Aicardi-Goutieres syndrome, (MIM:	0	0,22	NA
	GCA (MIM: 607030), Grancalcin	+507,649/0	NA	0	0,12	Mouse: Increased resistance to endotoxic
	KCNH7 (MIM: 608169), Potassium channel	+560,216/+1	NA	0,98	0,9	Schizophrenia, bipolar spectrum disorder[4 5]
	TBR1 (MIM: 604616), T-box brain 1	-385,322/-1	NA	0,99	1	Intellectual disability possibly autosomal
	PSMD14 (MIM: 607173), Proteasome 26s	-399,475/-1	NA	0,79	0,98	NA
	TANK (MIM: 603893), Traf family member-	-574,971/-1	NA	0,94	0,77	Mouse: Increased mortality due to
	CCDC67 (MIM: 617148), Coiled-coil	+86,326/0	NA	0	0,08	NA
	SMCO4 (MIM: 609477), Single-pass membrane	+234,827/0	NA	0	0,29	NA
	TAF1D (MIM: 612823), TATA box-binding	+486,303/0	NA	0,01	0,05	NA
	KIAA1731 (MIM: 617728), Centrosomal	+417,994/0	NA	0	0,11	NA

46,XY,t(2;11)(q24.2;q21), chr11	*C11orf54* (MIM:615810),	+497,946/0	NA	0,01	0,07	NA
	MED17 (MIM: 603810), Mediator complex	+540,582/0	Microcephaly, postnatal	0	0,26	NA
	HEPHL1 (NA), Hephaestin Like 1	+777,716/+1	NA	0	0,12	NA
	PANX1 (MIM: 608420), Pannexin 1	+885,283 /+1	NA	0	0,13	Intellectual disability, autosomal recessive[8]
	FOLR4 (MIM: 615737), Folate receptor 4	+1,062,066/+1	NA	0	0,12	Mouse: female infertility
	GPR83 (MIM: 605569), G Protein-Coupled Receptor 83	+1,134,090/+1	NA	0,03	0,15	Mouse: delayed spatial learning, stress provoked anxiety
	MRE11A (MIM: 600814), Meiotic Recombination 11 Homolog A	+1,175,879/+1	Ataxia-telangiectasia-like disorder (MIM: 604391), AR	0	0,25	NA
	ANKRD49 (NA), Ankyrin Repeat Domain 49	+1,250,362/+1	NA	0,17	0,08	NA
	FUT4 (MIM: 104230), Fucosyltransferase 4	+1,300,245/+1	NA	0,06	0,46	Mouse: increase in neutrophils, eosinophils and monocytes
	PIWIL4 (MIM: 610315), Piwi Like RNA-	+1,323,798/+1	NA	0	0,1	Mouse: Male infertility
	SLC36A4 (MIM: 613760), Solute Carrier Family 36 Member 4	- 45,687 /0	NA	0	0,14	NA
	MTNR1B (MIM: 600804), Melatonin Receptor 1B	-258,585/0	Susceptibility to Diabetes mellitus, type 2 (MIM: 125853), AD	0,01	0,11	NA
	FAT3 (MIM: 612483), FAT Atypical Cadherin 3	-347,199/-1	NA	1	0,54	Hirschsprung disease, joint contribution[9]

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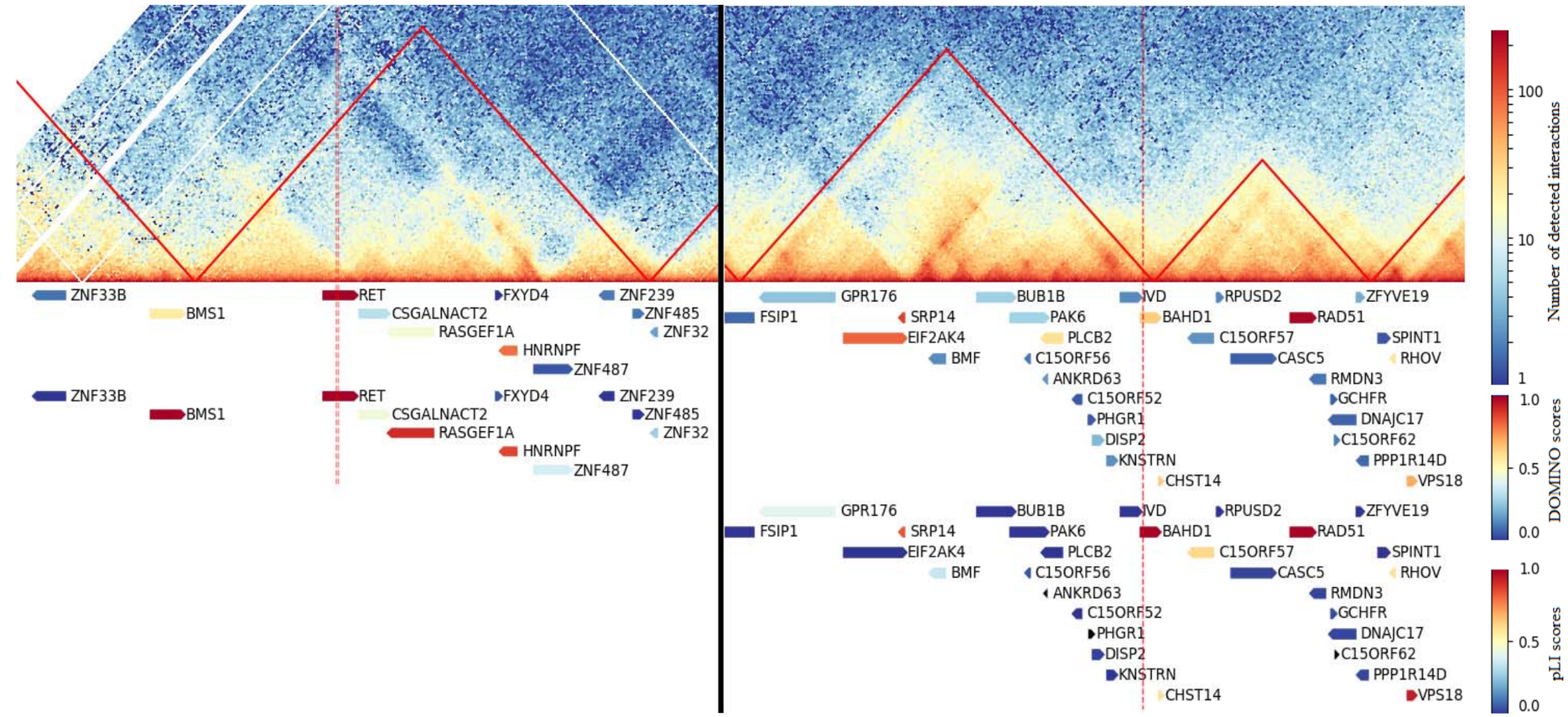
9. Luzon-Toro B, Gui H, Ruiz-Ferrer M, Sze-Man Tang C, Fernandez RM, Sham PC, Torroglosa A, Kwong-Hang Tam P, Espino-Paisan L, Cherny SS, Bleda M, Enguix-Riego Mdel V, Dopazo J, Antinolo G, Garcia-Barcelo MM, Borrego S. Exome sequencing reveals a high genetic heterogeneity on familial Hirschsprung disease. Sci Rep 2015;5:16473. doi: 10.1038/srep16473

46,XX,t(10;15)(q11.21;q15.1), chr10

46,XX,t(10;15)(q11.21;q15.1), chr15



Breakpoint	**Gene**	**Distance from breakpoint (bp)/domain**	**OMIM#, disease, inheritance**	**pLI**	**DOMINO**	**Comments** (HGMD, mouse phenotype)
46,XX,t(10;15)(q11.21;q15.1), chr10	*CSGALNACT2* (MIM: 616616), Chondroitin Sulfate N-Acetylgalactosaminyltransferase 2	+39,782/0	NA	0,45	0,32	Mouse: no abnormal phenotype
	RASGEF1A (MIM: 614531), RasGEF Domain Family Member 1A	+95,831/0	NA	0,91	0,46	NA
	HNRNPF (MIM: 601037), Heterogeneous Nuclear Ribonucleoprotein F	+286,913/0	NA	0,86	0,8	NA
	ZNF487 (NA), Zinc Finger Protein 487	+338,130/0	NA	0,38	0,05	NA
	ZNF239 (MIM: 601069), Zinc Finger Protein 239	+457,640/0	NA	0	0,11	NA
	ZNF485 (NA), Zinc Finger Protein 485	+ 507,703 /0	NA	0	0,1	NA
	ZNF32 (MIM: 194539), Zinc Finger Protein 32	+ 530,113/0	NA	0,28	0,18	NA

	BMS1 (MIM: 611448), BMS1, Ribosome Biogenesis Factor	- 268,397/-1	Aplasia cutis congenita, nonsyndromic (MIM: 107600), AD	1	0,56	NA
	ZNF33B (MIM: 194522), Zinc Finger Protein 33B	- 464,790/-1	NA	0	0,1	NA
46,XX,t(10;15)(q11.21;q15.1), chr15	*CHST14* (MIM: 608429), Carbohydrate Sulfotransferase 14	+ 27,297/0	Ehlers-Danlos syndrome,	0,59	0,62	NA
	C15orf57 (NA), Chromosome 15 open reading frame 57	+ 85,019/+1	NA	0,61	0,15	NA
	CASC5 (MIM: 609173), Kinetochore Scaffold 1	+150,355/+1	Microcephaly (MIM: 604321), AR	0,02	0,07	NA
	DNAJC17 (MIM: 616844), DnaJ Heat Shock Protein Family (Hsp40) Member C17	+324,204/+1	NA	0,03	0,08	Retinitis pigmentosa with hypogammaglobulinaemia, autosomal recessive[10]
	RPUSD2 (NA), RNA Pseudouridylate Synthase Domain Containing 2	+125,636/+1	NA	0	0,13	NA
	RAD51 (MIM: 179617), RAD51 Recombinase	+251,109/+1	Fanconi anemia (MIM: 617244),	0,99	0,99	NA
	RMDN3 (MIM: 611873), Regulator Of Microtubule Dynamics 3	+292,219/+1	NA	0,02	0,1	Mouse: no abnormal phenotype
	GCHFR (MIM: 602437), GTP Cyclohydrolase I Feedback Regulator	+320,355/+1	NA	0,04	0,07	NA
	PPP1R14D (MIM: 613256), Protein Phosphatase 1 Regulatory	+371,787/+1	NA	0,03	0,08	Mouse: Heart defects
	ZFYVE19 (NA), Zinc Finger FYVE-Type Containing 19	+363,421/+1	NA	0	0,2	NA
	SPINT1 (MIM: 605123), Serine Peptidase Inhibitor, Kunitz Type 1	+400,353/+1	NA	0	0,05	Mouse: Embryonic lethality, impaired growth development
	RHOV (NA), Ras Homolog Family Member V	+428,549/+1	NA	0,59	0,58	NA
	DLL4 (MIM: 605185), Delta Like Canonical Notch Ligand 4	+485,675/+1	Adams-Oliver syndrome (MIM: 616589), AD	0,98	1	NA
	CHAC1 (MIM: 614587), ChaC Glutathione Specific Gamma-Glutamylcyclotransferase 1	+509,297/+1	NA	0	0,17	NA
	INO80 (MIM: 610169), INO80 Complex Subunit	+535,215/+1	NA	1	1	Intellectual disability[11]

VPS18 (MIM: 608551), VPS18, CORVET/HOPS Core Subunit	+450,765/+1	NA	0,94	0,69	Mouse: impaired neuron migration and neurodegeneration
IVD (MIM: 607036), Isovaleryl-CoA Dehydrogenase	-7,720 /-1	Isovaleric acidemia (MIM: 243500), AR	0	0,14	NA
KNSTRN (MIM: 614718), Kinetochore Localized Astrin (SPAG5) Binding Protein	-49,419/-1	NA	0	0,15	Mouse: reduced male fertility
C15orf52 (NA), Chromosome 15 open reading frame 52	-102,698/-1	NA	0	0,06	NA
DISP2 (MIM: 607503), Dispatched RND Transporter Family Member 2	-72,609/-1	NA	0,02	0,23	NA
PLCB2 (MIM: 604114), Phospholipase C Beta 2	-135,730/-1	Platelet PLC beta-2 deficiency	0	0,59	NA
PAK6 (MIM: 608110), P21 (RAC1) Activated Kinase 6	-166,178/-1	NA	0	0,29	Mouse: no abnormal phenotype
C15orf56 (NA), Chromosome 15 open reading frame 56	-102,698/-1	NA	0,05	0,1	NA
BUB1B (MIM: 602860), BUB1 Mitotic Checkpoint Serine/Threonine Kinase B	-222,529 /-1	Mosaic variegated aneuploidy syndrome (MIM: 257300), AR	0	0,28	NA
BMF (MIM: 606266), Bcl2 Modifying Factor	-334,773/-1	NA	0,35	0,14	Mouse: Immunological disorders
EIF2AK4 (MIM: 609280), Eukaryotic Translation Initiation Factor 2 Alpha Kinase 4	-408,069/-1	Pulmonary venoocclusive disease (MIM: 234810), AR	0	0,81	NA
SRP14 (MIM: 600708), Signal Recognition Particle 14	-404,477/-1	NA	0,84	0,87	NA
GPR176 (MIM: 612183), G Protein-Coupled Receptor 176	-522,773/-1	NA	0,41	0,25	NA
FSIP1 (MIM: 615795), Fibrous Sheath Interacting Protein 1	-660,835/-1	NA	0	0,08	NA

10. Patel N, Aldahmesh MA, Alkuraya H, Anazi S, Alsharif H, Khan AO, Sunker A, Al-Mohsen S, Abboud EB, Nowilaty SR, Alowain M, Al-Zaidan H, Al-Saud B, Alasmari A, Abdel-Salam GM, Abouelhoda M, Abdulwahab FM, Ibrahim N, Naim E, Al-Younes B, A EA, AlIssa A, Hashem M, Buzovetsky O, Xiong Y, Monies D, Altassan N, Shaheen R, Al-Hazzaa SA, Alkuraya FS. Expanding the clinical, allelic, and locus heterogeneity of retinal dystrophies. *Genet Med* 2016;18(6):554-62. doi: 10.1038/gim.2015.127

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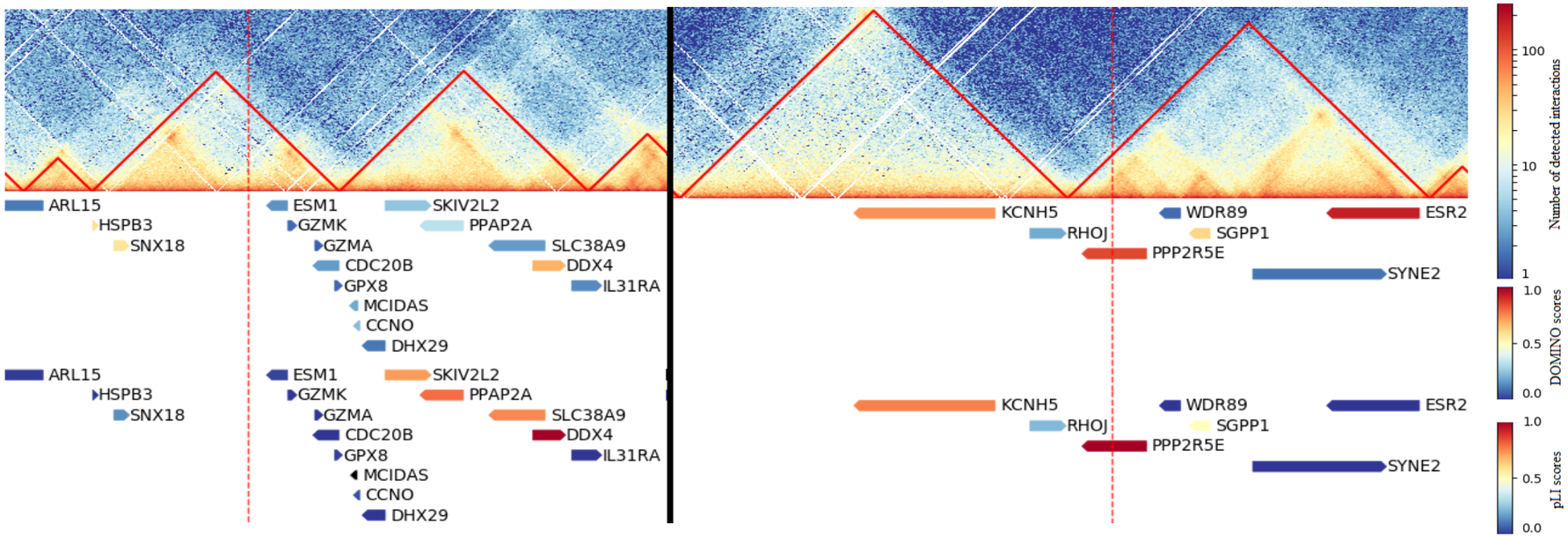
46,XX,t(5,14)(q11.2;q23.2),chr5

46,XX,t(5,14)(q11.2;q23.2),chr14



Breakpoint	**Gene**	**Distance from breakpoint (bp)/domain**	**OMIM#, disease, inheritance**	**pLI**	**DOMINO**	**Comments** (HGMD, mouse phenotype)
46,XX,t(5,14)(q11.2;q23.2),chr5	*ESM1* (MIM: 601521), Endothelial Cell Specific Molecule 1	+70,215/0	NA	0,03	0,15	NA
	GZMK (MIM: 600784), Granzyme K	+116,604/0	NA	0	0,08	NA
	GZMA (MIM: 140050), Granzyme A	+194,999/0	NA	0	0,07	Mouse: defective CTL cytolysis
	CDC20B (MIM: NA), Cell Division Cycle 20B	+205,322/0	NA	0	0,17	NA
	GPX8 (MIM: 617172), Glutathione Peroxidase 8	+252,469/0	NA	0,02	0,08	Mouse: chronic dermatitis, hyperkeratosis
	CCNO (MIM: 607752), Cyclin O	+323,503/0	Ciliary dyskinesia (MIM: 615872), AR	0,04	0,24	NA
	DHX29 (MIM: 612720), DExH-Box Helicase 29	+348,596/0	NA	0	0,11	Mouse: Embryonic lethality
	SKIV2L2 (NA), Ski2 Like RNA Helicase 2	+400,111/+1	NA	0,73	0,26	NA
	PPAP2A (MIM: 607124), Phospholipid Phosphatase 1	+517,205/+1	NA	0,8	0,32	NA
	SLC38A9 (MIM: 616203), Solute Carrier Family 38 Member 9	+718,196/+1	NA	0,76	0,17	NA

	DDX4 (MIM: 605281), DEAD-Box Helicase 4	+830,368/+1	NA	1	0,69	Mouse: Male infertility
	IL31RA (MIM: 609510), Interleukin 31 Receptor A	+944,042/+1	Amyloidosis (MIM: 613955), AD	0	0,14	NA
	SNX18 (NA), Sorting Nexin 18	-361,067/0	NA	0,15	0,58	NA
	HSPB3 (MIM: 604624), Heat Shock Protein Family B (Small) Member 3	-451,275/-1	Neuronopathy (MIM: 613376), AD	0	0,6	NA
	ARL15 (NA), ADP Ribosylation Factor Like	-337,078/-1	NA	0,01	0,11	Diaphragmatic hernia[12]
46,XX,t(5,14)(q11.2;q23.2),chr14	RHOJ (MIM: 607653), Ras Homolog Family Member J	-148,970/0	NA	0,23	0,2	Mouse: delayed growth in retina
	WDR89 (NA), WD Repeat Domain 89	+154,853/+1	NA	0	0,08	Mouse: no abnormal phenotype
	SGPP1 (MIM: 612826), Sphingosine-1-Phosphate Phosphatase 1	+242,028/+1	NA	0,5	0,62	Mouse: abnormal keratinocyte differentiation, postnatal lethality
	SYNE2 (MIM: 608442), Spectrin Repeat Containing Nuclear Envelope Protein 2	+410,779/+1	Emery-Dreifuss muscular dystrophy (MIM: 612999), AD	0	0,1	NA
	ESR2 (MIM: 601663), Estrogen Receptor 2	+642,046/+1	NA	0	0,94	Mouse: Neural, behavior, glucose homeostasis, cardiac, and ion channel abnormalities and myeloproliferative disorders
	KCNH5 (MIM: 605716), Potassium Voltage-Gated Channel Subfamily H Member 5	-340,152/-1	NA	0,77	0,74	Epileptic encephalopathy, autosomal dominant[13]

12. Longoni M, High FA, Qi H, Joy MP, Hila R, Coletti CM, Wynn J, Loscertales M, Shan L, Bult CJ, Wilson JM, Shen Y, Chung WK, Donahoe PK. Genome-wide enrichment of damaging de novo variants in patients with isolated and complex congenital diaphragmatic hernia. *Hum Genet* 2017;136(6):679-91. doi: 10.1007/s00439-017-1774-y

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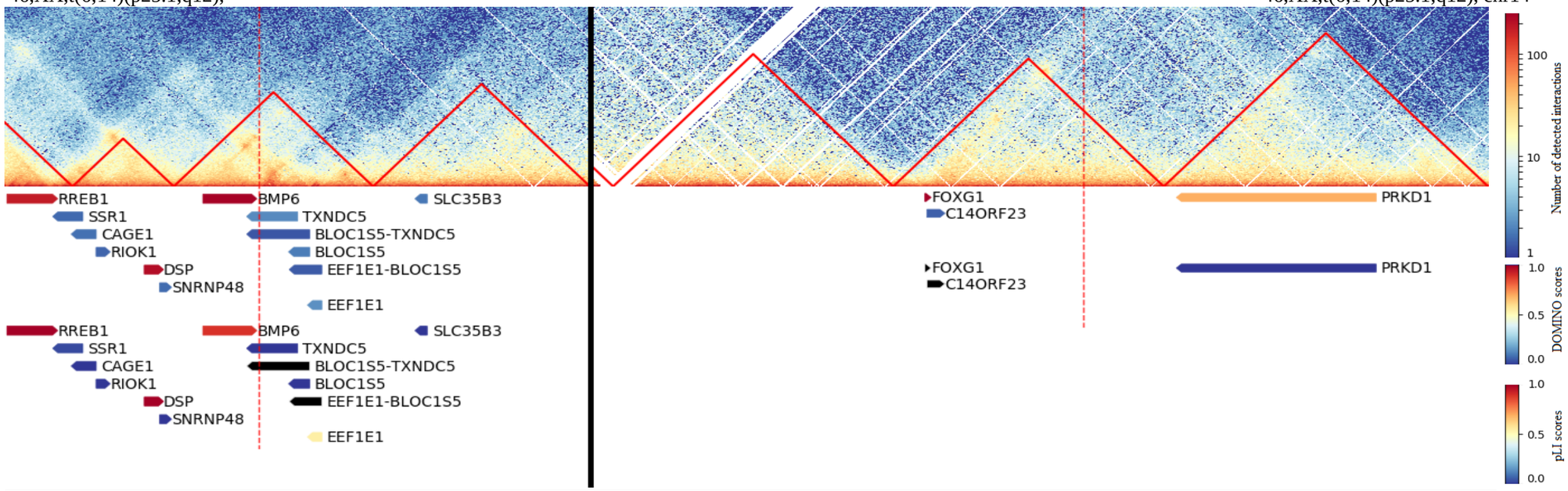
46,XX,t(6;14)(p25.1;q12),

46,XX,t(6;14)(p25.1;q12), chr14



Breakpoint	Gene	Distance from breakpoint (bp)/domain	OMIM#, disease, inheritance	pLI	DOMINO	**Comments** (HGMD, mouse phenotype)
46,XX,t(6;14)(p25.1;q12), chr6	*BLOC1S5* (MIM: 607289), Biogenesis Of Lysosomal Organelles Complex 1 Subunit 5	+110,367/0	NA	0	0,12	Mouse: Hermansky-Pudlak Syndrome
	EEF1E1 (MIM: 609206), Eukaryotic Translation Elongation Factor 1 Epsilon 1	+170,160/0	NA	0,55	0,15	Mouse: Embryonic lethality
	SLC35B3 (MIM: 610845), Solute Carrier Family 35 Member B3	+509,868/+1	NA	0	0,11	NA
	BMP6 (MIM: 112266), Bone Morphogenetic Protein 6	-21,778/0	NA	0,9	0,99	Iron overload , autosomal dominant[14]
	SNRNP48 (NA), Small Nuclear Ribonucleoprotein U11/U12 Subunit 48	-291,233/-1	NA	0	0,09	NA
	DSP (MIM: 125647), Desmoplakin	-316,483 /-1	Arrhythmogenic right ventricular dysplasia (MIM: 607450), AD, Dilated cardiomyopathy with woolly hair (MIM: 615821) AD, Keratosis palmoplantaris striata (MIM: 612908) AD	1	0,97	NA
	RIOK1 (MIM: 617753), RIO Kinase 1	-485,163/-1	NA	0	0,08	NA
	CAGE1 (MIM: 608304), Cancer Antigen 1	-513,457/-1	NA	0	0,09	NA

	SSR1 (MIM: 600868), Signal Sequence Receptor Subunit 1	-555,754/-2	NA	0,03	0,08	Mouse: neonatal lethality, impaired cardiac development
	RREB1 (MIM: 602209), Ras Responsive Element Binding	-651,220/-2	NA	1	0,94	Bipolar disorder, autosomal dominant[15] Congenital heart
46,XX,t(6;14)(p25.1;q12), chr14	*PRKD1* (MIM: 605435), Protein Kinase D1	+310,163/+1	Congenital heart defects and ectodermal dysplasia (MIM: 617364), AD	0	0,7	NA
	FOXG1 (MIM: 164874)	-/0	Rett syndrome (MIM: 613454), AD	NA	1	NA

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