Supplementary Material

FAN1, a DNA Repair Nuclease, as a Modifier of Repeat Expansion Disorders

Supplementary Table 1. *FAN1* mutations associated with KIN

Sr No.	Nucleotide alteration	Protein Change	Exon/Intron	State	Reference
1	c.1234+2T>A	Splice site	2	Heterozygous	[1]
	c.2036_7delGA	p.R679Tfs*5	7		
2	c.1234+2T>A	Splice site	2	Heterozygous	[1]
	c.2245C>T	p.R749*	9		
3	c.1375+1G>A	Splice site	3	Heterozygous	[1]
	c.2616delA	p.R873T*17	12		
4	c.1606c>T	p.R536*	5	Heterozygous	[1]
	c.2786A>C	p.Q929P	12		
5	c.1606c>T	p.R536*	5	Heterozygous	[1]
	c.2878G>A	p.D960N	13		
6	c.2120G>A	p.W707*	8	Homozygous	[1]
7	c.2616delA	p.D873T*17	12	Homozygous	
8	c.2611T>C	p.C871R	12	Heterozygous	[1]
	c.2878G>A	p.D960N	13		
9	c.2774_5delTT	p.L925Pfs*25	12	Heterozygous	[1]
	c.2810G>A	p.G937D	13		
10	c.1102C>T	p.N368*	2	Heterozygous	[2]
	c.2616delA	p.D873Tfs*17	12		
12	c.2590C>T	p.E864*	12	Heterozygous	[3]
	c.2774_5delTT	p.L925Pfs*25	12		
13	c.1899del	p.C633Wfs*9	6	Homozygous	[4]
14	c.1520G>A	p.R507H	4	Heterozygous	[5]
15	c.1369C>T	p.Q457*	4	Heterozygous	[6]
	c.1356T>G	p.N452K	4		

Nomenclature for variants are used as described in [7].

Supplementary Table 2. *FAN1* Variants
(Summary of *FAN1* variants in various diseases. These variants do not necessarily lead to disease)

Sr No.	Variant	Amino acid change	Diseases	Reference
Missense mutations				
1	rs148404807	p.M50R	CRC, Pancreatic Cancer	[8]
2	rs148404807	p.M50T	ASD, SCZ	[9]
3	rs761776412	p.D140Y	CRC	[10]
4	rs771206220	p.P340S	CRC	[10]
5	rs151322829	p.R377W	HD, Breast cancer	[11,12]
6	rs201153099	p.L395P	ASD	[9]
7	rs150393409	p.R507H	Breast cancer, ASD, SCZ, HD	[9,11,12]
8		p.K505I	ASD	[9]
9	rs377418523	p.R591W	CRC	[10]
10	rs144081053	p.P654L	ASD	[9]
11	rs200818425	p.R658Q	ASD	[9]
12	rs145610507	p.K794R	ASD	[9]
Nonsense mutation				
13		p.S18*	Breast and ovarian cancer	[13]
14		p.C47*	CRC	[13]
15		p.S258*	Breast and ovarian cancer	[13]
16		p.S466*	Breast and ovarian cancer	[13]
17		p.R536*	KIN, Breast and ovarian cancer	[1,13]
18		p.R706*	Breast and ovarian cancer	[13]
19		p.R710*	Breast and ovarian cancer	[13]
20		p.R952*	Breast, ovarian and other cancer	[10,13]
Frame shift mutation				
21		p.K11G fs*45	Breast and ovarian cancer	[13]
22		p.K11S fs*45	Breast and ovarian cancer	[13]
23		p.T187S fs*	Breast and ovarian cancer	[13]
24		p.V308Cfs*5	Breast, ovarian and other cancer	[13]
25		p.E320Gfs*28	CRC	[13]
26		p.S323Ifs*3	Breast and ovarian cancer	[13]
27		p.R679T fs*5	Breast and ovarian cancer	[13]
28		p.D873TFS*17	CRC	[2,13]
	Non-Coding			
29	rs4779794	Non-Coding	BPD/SCZ/MDD	[14]
30	rs7171212	Non-Coding	BPD/SCZ	[14]
31	rs4779796	Non-Coding	SCZ	[14]
	CNV			
32		15q13.2	ASD, SCZ and KIN	[9,15,16]
33		15q13.3	ASD, SCZ and KIN	[9,15,16]

ASD, Autism spectrum disorder; SCZ, Schizophrenia; KIN, Karyomegalic interstitial nephritis; CRC, Colorectal cancer; BPD, Bipolar disorder; MDD, Major depressive disorder; HD, Huntington's disease.

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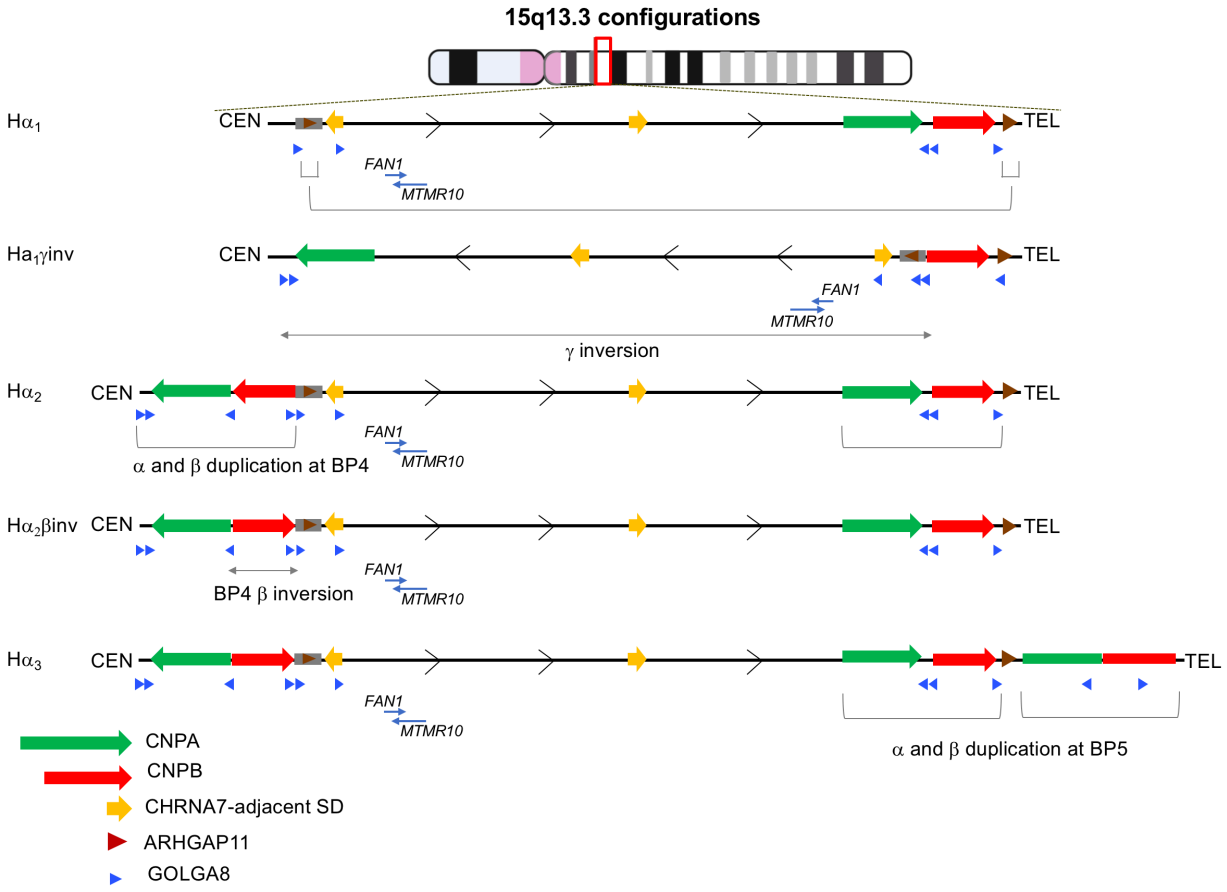

Supplementary Figure 1. Different configurations of the human 15q13.3 region. See text for detail.