The American Journal of Human Genetics

Supplemental Data

Joubert Syndrome in French Canadians and Identification of Mutations in *CEP104*

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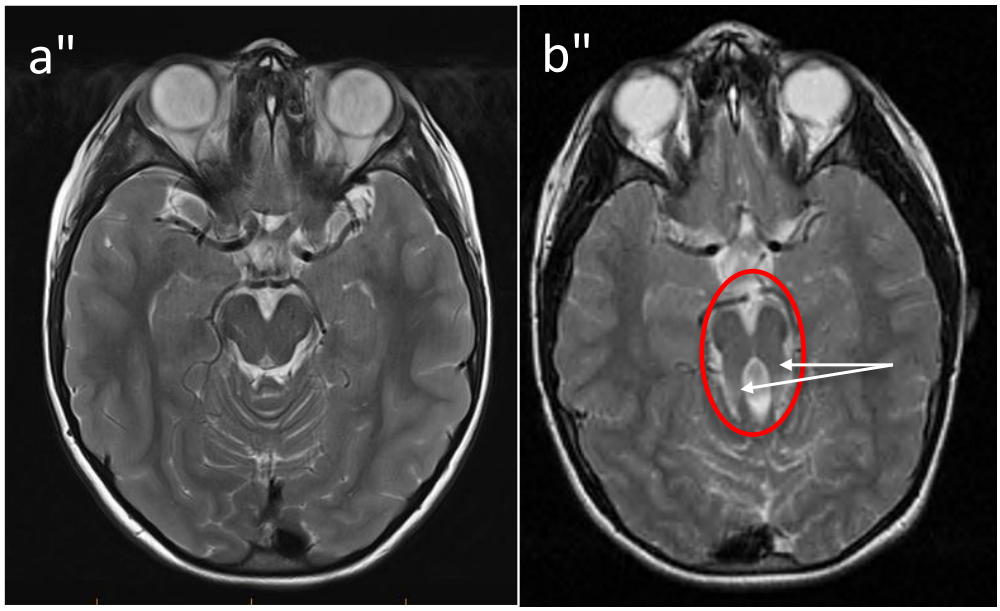

Figure S1. Molar Tooth Sign on MRI. Axial T2 brain MRI of a control individual (a) and patient with JBTS (b). Note the molar tooth sign (red circle), thickened and abnormally oriented superior cerebellar peduncles (white arrows) and deepened interpeduncular fossa.

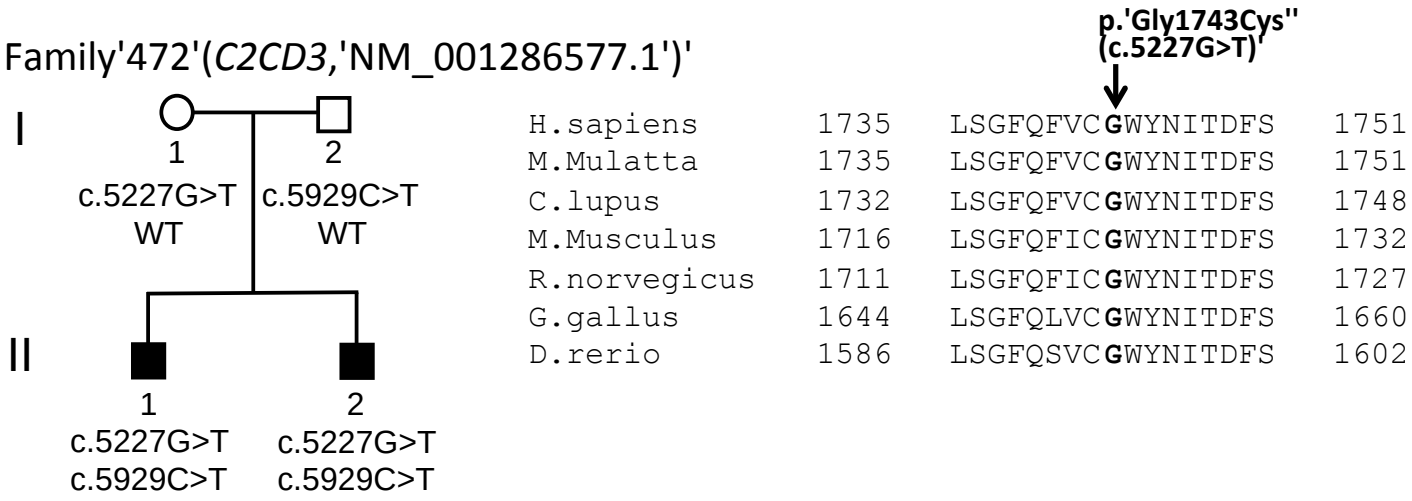

Figure S2. Segregation of mutations in *C2CD3*. The panel on the right shows conservation of the affected amino acid in C2CD3. Amino acid alignments were generated using homologene (NCBI).

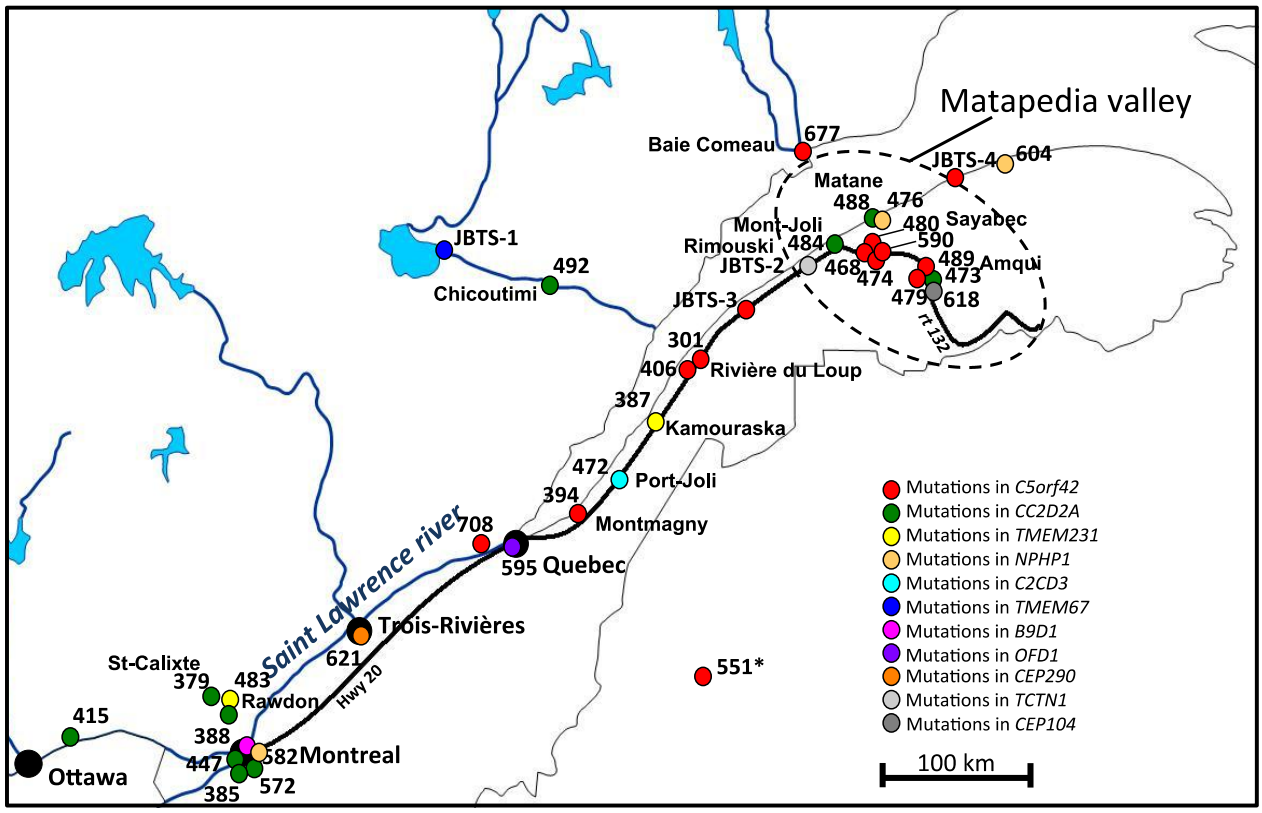

Figure S3. Map of Quebec showing the geographic distribution and the genetic heterogeneity of FC families with JBTS. *Note that this individual is of FC ancestry, but born and living in the Unites States of America.

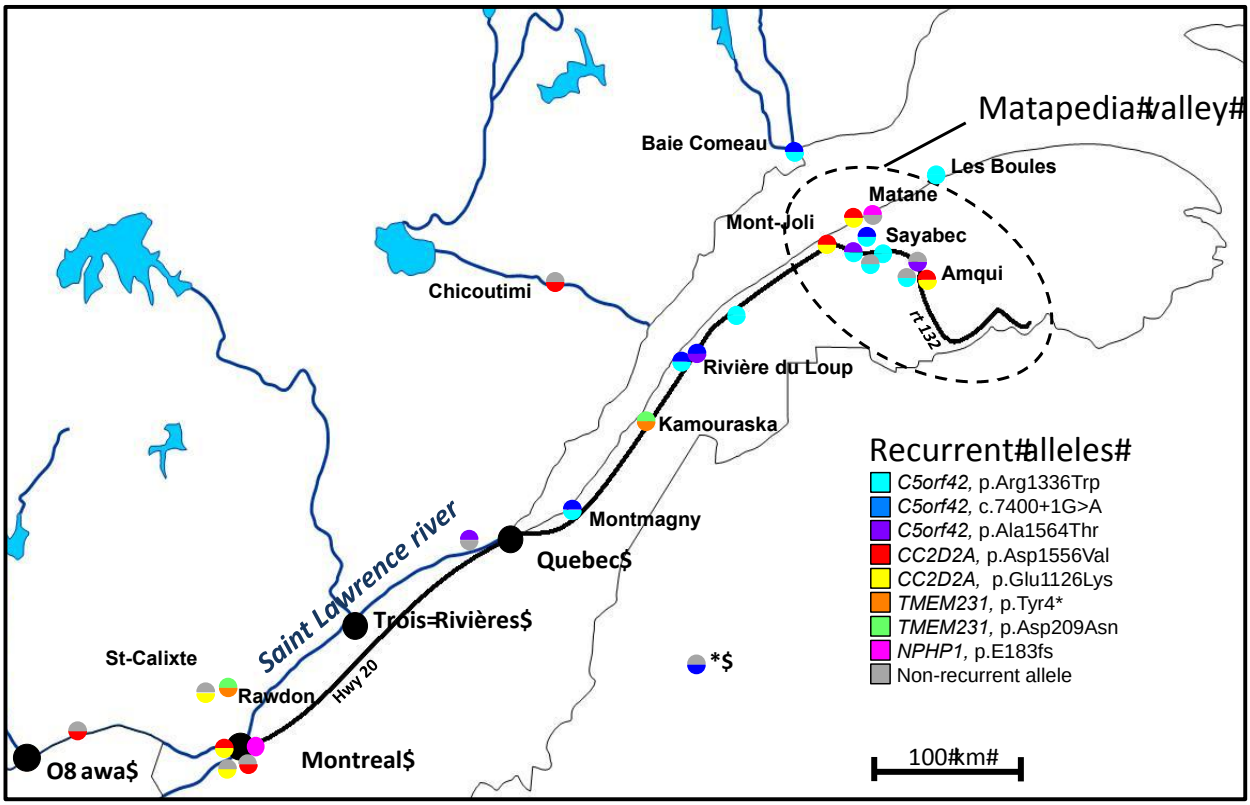

Figure S4. Map of Quebec showing the geographic distribution of the recurrent FC JBTS alleles. *Note that this individual is of FC ancestry, but born and living in the Unites States of America.

Table S2. Rare homozygous and potentially compound heterozygous variants in individual 1763.618

Chr	Position (hg 19)	Ref Allele	Alt Allele	Variant	dbSNP	1KG (MAF)	EVS (MAF)	ExAC (MAF)	SIFT score	PolyPhen-2	Zygosity
1	3756170	A	G	CEP104:NM_014704:c.735+2T>C	NR	NR	NR	NR	NA	NA	Homoz.
8	145024570	C	T	PLEC:NM_201380:c.G305A:p.R102H	rs200335928	NR	0.0048	0.0026	0.00 (D)	0.93 (PD)	Homoz.
2	186653963	G	C	FSIP2:NM_173651:c.G2367C:p.L789F	NR	NR	NR	NR	0.00 (D)	0.99 (PD)	Het.
2	186660153	C	A	FSIP2:NM_173651:c.C8557A:p.P2853T	NR	NR	NR	NR	0.16 (T)	NA	Het.
7	28995453	T	C	TRIL:NM_014817:c.A2209G:p.T737A	rs552268645	NR	NR	0.0008	NA	NA	Het.
7	28995666	C	T	TRIL:NM_014817:c.G1996A:p.G666R	rs562686571	NR	NR	0.0009	NA	0.04 (B)	Het.
7	149482585	C	T	SSPO:NM_198455:c.C3001T:p.R1001W	rs199648588	NR	0.0025	0.003	0.01 (D)	1.0 (PD)	Het.
7	149502579	C	T	SSPO:NM_198455:c.C8392T:p.R2798C	rs181269877	0.0041	0.0037	0.006	NA	1.0 (PD)	Het.
8	145732114	G	C	GPT:NM_005309:c.G1288C:p.E430Q	rs141505249	0.0027	0.0016	0.004	0.33 (T)	0.03 (B)	Het.
8	145732180	G	C	GPT:NM_005309:c.G1354C:p.V452L	rs147998249	0.0027	0.0016	0.004	0.01 (D)	0.2 (B)	Het.
9	137676938	A	T	COL5A1:NM_000093:c.A2588T:p.E863V	rs139788610	NR	0.00077	0.0004	0.00 (D)	0.986 (PD)	Het.
9	137701080	G	A	COL5A1:NM_000093:c.G3418A:p.V1140M	rs149616140	NR	0.00046	0.0008	0.03 (D)	0.99 (PD)	Het.
10	91498041	C	T	KIF20B:NM_016195:c.C3323T:p.A1108V	rs117564945	0.0009	0.0031	0.0044	0.31 (T)	0.0 (B)	Het.
10	91520368	T	A	KIF20B:NM_016195:c.T4646A:p.F1549Y	rs117258675	0.0009	0.003	0.0045	0.15 (T)	0.302 (B)	Het.
17	18881091	G	A	FAM83G:NM_001039999:c.C1888T:p.R630W	rs201046878	NR	0.0023	0.0026	0.00 (D)	0.97 (PD)	Het.
17	18907165	G	A	FAM83G:NM_001039999:c.C190T:p.L64F	rs371100508	NR	0.00008	0.00002	0.01 (D)	0.926 (PD)	Het.
17	73237124	T	C	GGA3:NM_138619:c.A961G:p.S321G	rs146877619	0.0009	0.0025	0.0021	0.31 (T)	0.0 (B)	Het.
17	73239557	T	C	GGA3:NM_138619:c.A395G:p.K132R	rs117805695	0.0014	0.0026	0.0022	0.1 (T)	0.001 (B)	Het.

B, benign; D, damaging; NR, not reported; NA, not available; PD, probably damaging; T, tolerated; Het., heterozygous; Homoz., homozygous