American Journal of Human Genetics, Volume *90*

Supplemental Data

RAD51 Haploinsufficiency Causes

Congenital Mirror Movements in Humans

Christel Depienne, Delphine Bouteiller, Aurélie Méneret, Ségolène Billot, Sergiu Groppa, Stephan Klebe, Fanny Charbonnier-Beaupel, Jean-Christophe Corvol, Jean-Paul Saraiva, Norbert Brueggemann, Kailash Bhatia, Massimo Cincotta, Vanessa Brochard, Constance Flamand-Roze, Wassila Carpentier, Sabine Meunier, Yannick Marie, Marion Gaussen, Giovanni Stevanin, Rosine Wehrle, Marie Vidailhet, Christine Klein, Isabelle Dusart, Alexis Brice, and Emmanuel Roze

Table S1. Clinical features of patients with *RAD51* mutations

Family-Patient Gender/age	MM severity[a]	MM location	Associated disorders	Course [c]	Functional disability	Treatment of MM
A-3 F/81	0[b]	-	-	-	-	-
A-9 M/59	3	Hands Forearms	No	stable	Difficulties in fine bimanual activities Pain/cramp during sustained manual activities writing fatigability	No
A-11 F/56	0[b]	-	-	-	-	-
A-15 M/53	2	Hands Forearms	No	stable	Pain/cramp during sustained manual activities	No
A-17 M/52	2	Hands	No	stable	Pain/cramp during sustained manual activities	No
A-21 M/50	0[b]	-	-	-	-	-
A-37 M/39	0[b]	-	-	-	-	-
A-41 M/34	0[b]	-	-	-	-	-
A-53 M/27	3	Hands Forearms	No	stable	Difficulties in fine bimanual activities Pain/cramp during sustained manual activities	No
A-54 F/31	3	Hands Forearms	No	stable	Difficulties in fine bimanual activities Pain/cramp during sustained manual activities writing fatigability	No
A-56 M/33	2	Hands Forearms	No	Stable	Difficulties in fine bimanual activities Pain/cramp during sustained manual activities	No

A-65 F/17	3	Hands Forearms Feet	No	stable	Difficulties in fine bimanual activities Pain/cramp during sustained manual activities writing fatigability	No
A-85 F/14	0[b]	-	-	-	-	-
A-86 F/16	0[b]	-	-	-	-	-
A-87 F/20	0[b]	-	-	-	-	-
A-91 M/13	2	Hands Forearms	No	stable	Difficulties in fine bimanual activities Pain/cramp during sustained manual activities	No
B-1 F/58	2	Hands Forearms	No	stable	Difficulties in fine bimanual activities	No
B-2 M/31	2	Hands Forearms	No	stable	Difficulties in fine bimanual activities	No

[a] according to the Woods and Teuber MM severity scale [24]: (0. No MM; 1. Barely discernible but repetitive MM; 2. Either slight but sustained MM, or stronger, but briefer MM; 3. Strong and sustained repetitive MM; 4. MM equal to that observed in the intended hand
[b] asymptomatic carriers
[c] Course over decades from onset in infancy or early childhood

Table S2. Statistics of SNPs and indels detected by whole exome analysis in patients 54 and 91 from Family A

	SNP								Indels		
Ind	Total	Htz	Known	New	in CDS	New in CDS	Nonsense	Missense	Total	Htz	in CDS
54	32 390	18 871	30 202	2 188	16 640	1 069	66	7 699	1 911	1 164	241
91	33 648	20 182	31 558	2 090	17 259	950	60	8 002	1 948	1 187	225

The coding regions of the genome of patients 54 and 91 (Family A) were sequenced by Integragen SA (Evry, France). Genomic DNA was captured using Agilent in-solution enrichment methodology (SureSelect Human All Exon Kits Version 1, Agilent) with the biotinylated oligonucleotide probe library (Human All Exon v1 – 38 Mb, Agilent), followed by paired-end 75 b massively parallel sequencing on Illumina GAIIx. Image analysis and base calling were performed using Illumina Real Time Analysis (RTA) Pipeline version 1.8 with default parameters. Bioinformatic analysis of sequencing data was based on the Illumina pipeline (CASAVA1.7). CASAVA performs alignment of a sequencing run to a reference genome (hg19), calls the SNPs based on the allele calls and read depth, and detects variants (SNPs & Indels). The alignment algorithm used was ELANDv2 (performs multiseed and gapped alignments). Htz, variants at the heterozygous state.

Supplemental Reference

24. Woods, B.T., and Teuber, H.L. (1978). Mirror movements after childhood hemiparesis. Neurology *28*, 1152–1157.