

Table S3 Phenotypes in patients/subjects with increased copy number of *BHLHA9*

Patient			Ectrodactyly		Long bone deficiency		Phenotype
Family	Subject	Sex	Hands	Feet	Tibia	Femur	
1	II-1	M	+/+	+/+	+/+	+/-	GWC
2	II-1	F	+/+	+/-	+/-	-/-	SHFLD
	II-2	M	+/+	+/+	+/+	-/-	SHFLD
3	I-1	M	-/+	-/-	+/-	-/-	SHFLD
	II-2	F	+/+	-/+	-/-	-/-	SHFM
	II-3	F	+/-	-/-	-/-	-/-	SHFM
4	II-1	M	+/-	-/-	+/-	-/-	SHFLD
5	II-1	F	-/+	-/-	-/+	-/-	SHFLD
6	II-1	M	+/-	+/+	+/+	-/-	SHFLD
7	II-1	M	+/-	-/+	-/-	-/-	SHFM
	II-3	F	+/-	-/-	-/-	-/-	SHFM
	III-2	M	+/+	-/-	-/-	-/-	SHFM
	IV-1	F	+/+	-/-	-/-	-/-	SHFM
8	I-1	M	+/-	+/-	-/-	-/-	SHFM
	II-1	M	+/-	+/-	-/-	-/-	SHFM
9	I-1	M	+/+	-/-	-/-	-/-	SHFM
	II-2	M	+/+	-/-	-/-	-/-	SHFM
10	I-2	F	+/+	-/-	-/-	-/-	SHFM
	II-1	F	+/-	-/-	-/-	-/-	SHFM
	II-2	F	-/+	-/-	-/-	-/-	SHFM
11	II-1	F	+/-	-/-	-/-	-/-	SHFM
12	II-1	M	+/-	-/-	-/-	-/-	SHFM
	II-3	M	-/+	-/-	-/-	-/-	SHFM
13	II-3	F	+/+	-/-	-/-	-/-	SHFM
14	II-1	M	-/+	-/-	-/-	-/-	SHFM
15	II-2	M	+/-	-/-	-/-	-/-	SHFM
16	II-3	M	+/-	-/-	-/-	-/-	SHFM
17	II-2	F	+/-	-/-	-/-	-/-	SHFM
18	II-1	M	+/+	+/+	-/-	-/-	SHFM
19	II-2	F	+/+	-/-	-/-	-/-	SHFM
20	II-1	M	+/+	+/-	-/-	-/-	SHFM
21	II-1	F	+/+	-/-	-/-	-/-	SHFM
22	II-1	M	+/+	-/-	-/-	-/-	SHFM
23	II-1	M	-/-	+/-	+/+	+/-	GWC
	II-2	M	+/+	+/-	+/-	-/-	SHFLD
24	II-1	M	+/-	+/-	+/-	-/-	SHFLD
	III-1	M	+/+	+/-	+/+	-/-	SHFLD
25	III-1	M	+/+	+/+	+/+	-/-	SHFLD
	III-2	F	+/+	-/-	-/-	-/-	SHFM
26	II-2	M	-/+	-/-	+/+	-/-	SHFLD
27	I-2	F	+/+	+/+	-/-	-/-	SHFM
	II-3	M	-/+	-/+	-/-	-/-	SHFM

Right/Left.

Patient numbers correspond to those of Fig. 1.

Patient 3-II-2 has ventricular septal defect and aortic coarctation.

Patient 13-II-1 has hypospadias.

Patient 15-II-2 has polydactyly of the left foot.