

## **Supplementary Documents**

### **Supplementary Tables**

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Supplementary Figure 1. Full pedigree.

Supplementary Figure 2. Clinical and radiological features of hands and feet of the remaining family members with the chromosomal deletion.

Supplementary Figure 3. Quantitative real-time PCR analysis of *DYNC111* mRNA in osteoblasts of the proband, presented relative to *GAPDH* mRNA. *DYNC111* 5'-B denotes *DYNC111*'s mRNA level determined by a primer pair amplifying exons 11-12, which were 5' to the 5' breakpoint of the deletion. *DYNC111* 3'-B denotes *DYNC111*'s mRNA level determined by a primer pair amplifying exons 16-17, which were 3' to the 5' breakpoint of and were within the deletion.

**Supplementary Table 1.** Phenotype of the affected family members.

ID	Sex	Malformation			
		Left hand	Right hand	Left foot	Right foot
II-2	M	Cleft	None	Cleft	None
II-7	M	None	Cleft	Cleft	Cleft
II-11	M	Cleft	Cleft	Cleft	Cleft
III-4	M	Cutaneous syndactyly between 2 <sup>nd</sup> and 3 <sup>rd</sup> fingers	Cutaneous syndactyly between 2 <sup>nd</sup> and 3 <sup>rd</sup> fingers	None	None
III-22	M	None	Central polydactyly (an extra digit between 2 <sup>nd</sup> - 3 <sup>rd</sup> fingers was surgically removed)	None	None
III-30	F	None	None	Cutaneous between 1 <sup>st</sup> and 2 <sup>nd</sup> toes (post surgically separation)	None
IV-10	M	None	Cutaneous syndactyly between 3 <sup>rd</sup> -4 <sup>th</sup> fingers	None	Cleft
IV-30	M	Cleft and cutaneous syndactyly between 3 <sup>rd</sup> and 4 <sup>th</sup> fingers	Cleft	Cleft	Cleft

**Supplementary Table 2.** Taqman probes for RT-PCR

Gene	Probe	Exon boundary
<i>DYNC111</i>	Hs 00189392_m1	11-12
<i>DYNC111</i>	Hs 00920186_m1	16-17
<i>DSS1</i>	Hs 00428732_m1	2-3
<i>DLX6</i>	Hs 00231999_m1	1-2
<i>DLX5</i>	Hs 00193291_m1	1-2

**Supplementary Table 3.** Primers for genotyping and expression analyses

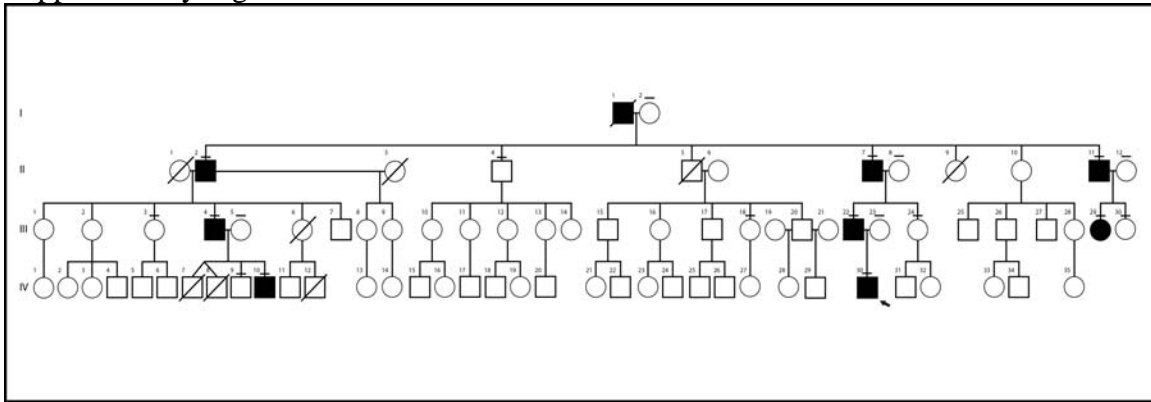
Primer name	Nucleotide sequence (5'>3')	Length (bp)	Product sizes (bp)	Annealing temperature (°C)
Primer sequences for deletion mutation analysis				
SHFM_del_F2	CCG-CCC-ATA-CAA-CCT-CAT-TT	20	2245	62
SHFM_del_R6	GAT-GTC-TGA-AGT-CAG-CAA-CC	20		
MLL3_Ex14_F	CTT-GTG-TAC-TTT-GGC-TTT-CAT-TCA-C	25	1188	62
MLL3_Ex14_R	GCT-CTA-TAT-TTA-CTG-CAG-CAC-AGA-AC	26		
Primer sequences for <i>DSS1</i> mutation analysis				
DSS1_5'UTR_F1	GTC-GGA-GGC-GGT-AGT-GAC	18	249	63
DSS1_5'UTR_R1	CCC-GAG-CAC-CCA-AGC-TAC	18		
DSS1_Ex2_F1	TTG-TGA-GTG-TTC-AGT-AGC-AAG-C	22	373	63
DSS1_Ex2_R1	TGC-AAT-TAC-ACA-AAG-TGT-TCA-TGT	24		
DSS1_3'UTR_F1	TGA-TGT-CTC-AGG-CAT-AGC-TCA	21	276	63
DSS1_3'UTR_R1	CAA-AAA-CAC-AAA-TAA-ATC-CAA-GCA	24		
Primer sequences for <i>DSS1</i> allelic expression analysis				
DSS1_5'UTR_F1	GTC-GGA-GGC-GGT-AGT-GAC	20	771	62
DSS1_3'UTR_R4	TGT-CAG-CAA-ATG-TTG-CCT-AAG	21		
Primer sequences for <i>DLX6</i> mutation analysis				
DLX6_5'UTR_F1	GCG-TCT-GCA-TCA-AGT-CTG-AA	20	931	63
DLX6_5'UTR_R2	GGG-CGT-CGA-GGA-AGT-GAG-CG	20		
DLX6_Ex2_F1	TAT-TGG-GCT-TTG-GGG-AGA-CT	20	382	65
DLX6_Ex2_R1	TAG-GCC-AAC-AAT-TCC-TCC-TG	20		
DLX6_3'UTR_F1	GAC-TTG-GCT-TGT-AGG-CGT-TG	20	1454	63
DLX6_3'UTR_R3	AGG-GAA-GTG-AGG-GAG-GAG-GG	20		
Primer sequences for <i>DLX6</i> allelic expression analysis				
DLX6_Ex2_F2	GAA-AAC-GGG-GAA-ATC-AGG-TT	20	1412	62
DLX6_3'UTR_R4	CGT-TTT-GTG-GCT-TTT-ACT-CC	20		
Primer sequences for <i>DLX5</i> mutation analysis				
DLX5_5'UTR_F1	GAT-GGT-CGC-CCA-CCT-CAT-TC	20	1031	65

DLX5_5'UTR_R1	TGT-GAC-CCC-CAA-TCT-ACC-AC	20		
DLX5_Ex2_F1	GGT-GGT-GAT-ATG-GGC-AAA-GA	20	390	65
DLX5_Ex2_R1	AGT-CCC-ATC-GAG-ACT-GAA-CC	20		
DLX5_3'UTR_F1	AGA-TCA-TGA-AAA-ACG-GGG-AG	20	405	65
DLX5_3'UTR_R1	CAC-ATG-AAT-CTT-TTT-CAG-TTT-TCC	24		
Primer sequences for <i>DLX5</i> allelic expression analysis				
DLX5_cDNA_F3	AGA-TAG-ACT-AAG-ACC-CCT-CCC-CAC-T	25	1450	64
DLX5_3'UTR_R1	CAC-ATG-AAT-CTT-TTT-CAG-TTT-TCC	24		
Primer sequences for <i>DYNC111</i> mutation analysis				
DYNC111_Ex1_F	CTT-TCC-AGG-TCC-TCT-GCT-CG	20	600	64
DYNC111_Ex1_R	CCT-AGT-CTC-TCG-GCT-CTG-C	19		
DYNC111_Ex2_F	CGA-GCT-CAG-TGG-AGG-AAT-GT	20	821	64
DYNC111_Ex2_R	GGA-GCC-TCT-GGA-CCA-AAT-GT	20		
DYNC111_Ex3_F	CCC-ACG-TGA-TAG-AAG-CAG-GA	20	597	64
DYNC111_Ex3_R	ACA-CGG-TGG-AGA-AGG-TTG-AC	20		
DYNC111_Ex4_F1	GTC-TGA-CAC-TGC-TCT-TCA-CCA-G	22	383	64
DYNC111_Ex4_R	GCC-CAC-TCA-AAG-AAG-GAT-GA	20		
DYNC111_Ex5_F	AGC-AGC-CAT-AGC-TCC-CGA-TA	20	587	64
DYNC111_Ex5_R	CAG-AAC-TTG-TGT-GCC-CCA-CT	20		
DYNC111_Ex6_F1	TTG-TGG-TTT-GTT-TGT-TTT-AGT-GC	23	713	64
DYNC111_Ex6_R1	TGG-TAA-GAG-GAA-ATC-AAC-AAG-AAA	24		
DYNC111_Ex7_F	CTG-GAG-CTG-GAT-GTA-CAG-GTG	21	883	64
DYNC111_Ex7_R	GAG-AGC-CAA-GGG-CAT-AGT-GAG	21		
DYNC111_Ex8_F	CTT-TTT-GTG-GGG-ACA-TCT-GC	20	619	64
DYNC111_Ex8_R	AGT-CTC-TAT-CAC-CTG-GAC-TGT-TCC	24		
DYNC111_Ex9_F	CCT-TGC-CAG-GTC-AGG-TTA-GA	20	752	64
DYNC111_Ex9_R	GGA-CCT-CAA-GGA-TGG-TCA-GAG	21		
DYNC111_Ex10_F	ACA-GGA-GAG-GAA-GGG-AGT-AGC-AC	23	618	64
DYNC111_Ex10_R	CAC-ACA-GTC-CCT-AAC-CAA-GC	20		
DYNC111_Ex11_F	AGT-AAC-CAT-TGG-TCA-GCC-TCT-C	22	587	64
DYNC111_Ex11_R	AAG-GAG-ATC-CTC-CCT-ACA-TGC	21		

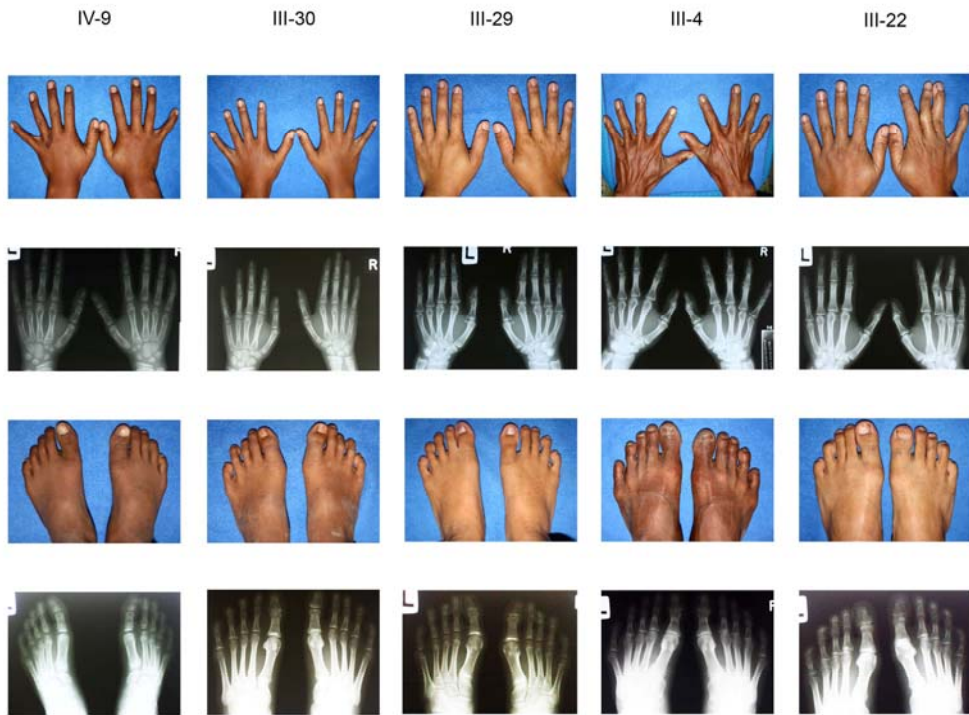
DYNC111_Ex12_F	CTG-ACT-GAG-CAA-GGT-GAA-GC	20	682	64
DYNC111_Ex12_R	CTC-CAA-CCT-CGT-CTG-CAT-CT	20		
DYNC111_Ex13_F	GGC-TGC-CCT-GCA-TGA-AAT-CT	20	906	64
DYNC111_Ex13_R	GGA-AGG-TGC-AAG-TGG-TCT-GT	20		
DYNC111_Ex14_F	CTG-CAT-CAG-CAA-TGA-TAG-GC	20	832	64
DYNC111_Ex14_R	GTA-GAT-ACC-ACC-AAG-CTC-CAG-AAC	24		
DYNC111_Ex15_F	GGT-CCA-CCT-GTC-TGG-AAT-GT	20	637	64
DYNC111_Ex15_R	CCA-CCG-TTT-AGG-GAT-GAA-GG	20		
DYNC111_Ex16_F	CCA-CTT-ACT-GCC-CGG-TGA-CT	20	660	64
DYNC111_Ex16_R	GGG-AAG-TGC-CAC-ACT-CCA-TC	20		
DYNC111_Ex17_F	AGG-CCC-TGC-TTG-ATC-CAA-AG	20	1167	64
DYNC111_Ex17_R	GTC-CCA-GCA-GAA-TTC-CAG-TC	20		
Primer sequences for <i>DYNC111</i> expression analysis				
DYNC111_Ex12_F1	CAG-CAT-CCC-GTG-TAC-TGT-GT	20	780	64
DYNC111_Ex17_R1	GTG-GCT-CAT-TTC-CAC-TAG-CG	20		

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Supplementary Figure 1.



Supplementary Figure 2.



Supplementary Figure 3.

