

Supplement data:

Table 1. Primers sequences information.

Primer name	Sequence
RUX2F2	GAGTACTGTGAGGTCACAAACCAC
RUX2R2	GCAACAAGATAGCATAATTAGGATC
RUX2F3	CAGACTCTGTTGGCCCATCAGAC
RUX2R3	GCAGGAGGTCTTGGAGGACGTC
RUX2F4	GTGGACCCTGAAACATTGGTC
RUX2R4	CAGAAGTTGATCCTCCAAGTTC
RUX2F5	GGACGATGATCTTGACTCTAAGC
RUX2R5	CTTAGTGATTGTGTGGCAAAGG
RUX2F6	GAATGCTGGCCACCAGATAC
RUX2R6	GCTGCAGAGTAGAGCATTAGTAG
RUX2F7	CTGAGTTTTGGGTTGCATGTTTC
RUX2R7	GCAGAGGAAATCCTGGCATAATAC
RUX2F8	CTGTCAGTTTTGGTTCTTCATC
RUX2R8	TGCACACTGCTTATAATCTTCAC
RUX2F9	AAGGATCAATGCTGTTAAGACTGC
RUX2R9	TATGGCTGCAAGATCATGACTG
Runxq2F	TGTGAGGTCACAAACCACATGA
Runxq2R	TGCTGTCCTCCTGGAGAAAGTT
Runxq3F	GTTTGAGGCTGGTCGTAGACAC
Runxq3R	AAGTTGTGGTCGAGAGGATGAA
Runxq4F	GGTCTGTTTGAAGTGGCATCAC
Runxq4R	AAAACAGGAAACACAGCAGCAA
Runxq5F	TCGGAGGGTTTCCAATTTAG

Runxq5R	TGGGGAGGATTTGTGAAGAC
Runxq6F	CAGTGATTTAGGGCGCATTC
Runxq6R	CCAGCGTCTATGCAAGTGAA
Runxq9F	CAGACCAGCAGCACTCCATATC
Runxq9R	ATCAGCGTCAACACCATCATTC
Hsa21-4F	CAATTCAGGTCAGGTGATAACTCAGTAA
Hsa21-4R	GCCAGGTTTAGAATGTTTGTCTAAGTC
44904142F	GAGTGGTAAAGTTAGGGCTTTTAGG
45403673R	CCAGAACTTGAATAGGAAGCC
45403507R	TGGCTGTGTTTCAGAGTCAGTT

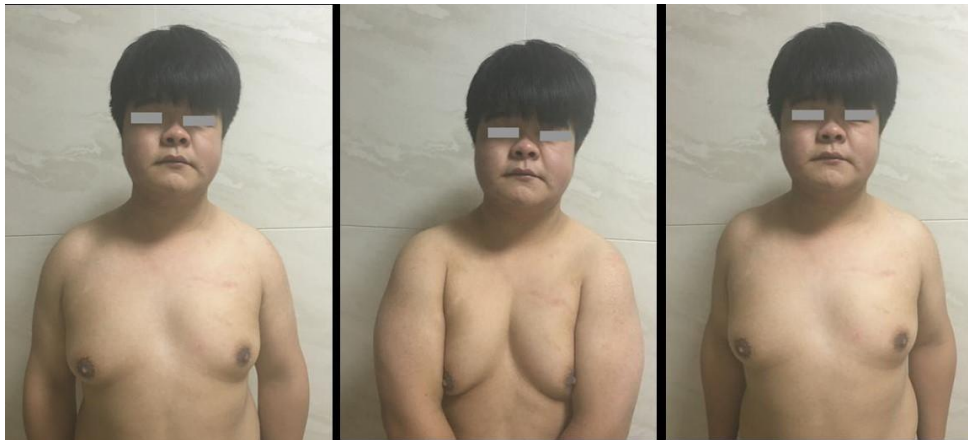


Figure 1. Shoulder mobility test confirmed that the proband's shoulders were not brought closer to meet in the middle of the body.

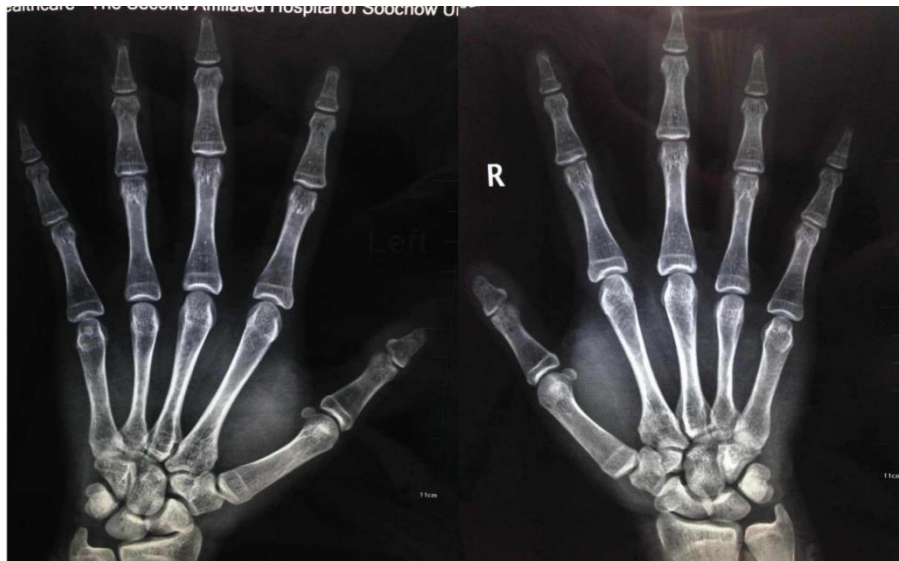


Figure 2. X-ray photos showed no brachydactyly or other hand malformation was found in the proband with CCD.

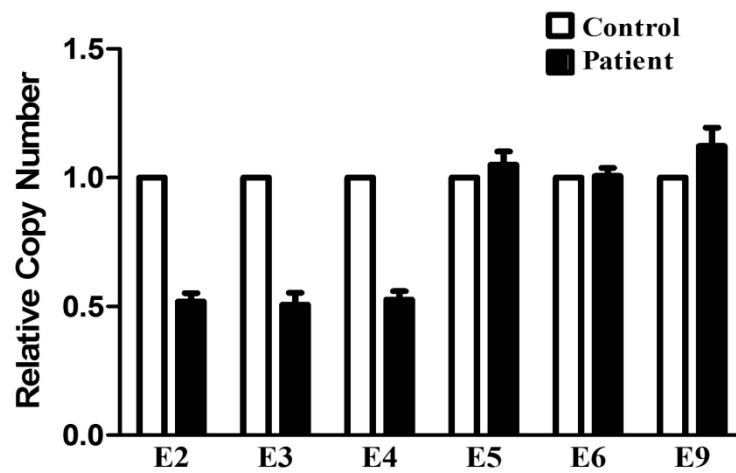


Figure 3. The deletion validation by q-PCR demonstrated that RCN was 0.5 for exons 2, 3 and 4 of *RUNX2* gene and RCN was 1.0 for exons 5, 6 and 9 of *RUNX2* gene.

AluSz6-291bp
chr6:44904413-44904703

AluSx-303bp
chr6:45403092-45403394

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Alignment statistics for match #1
NW Score      Identities  Gaps      Strand
188          219/303 (72%)  12/303 (3%)  Plus/Plus
AluSx  1      TTTTATCTTTTTTCTTTTTTGAGATGGAGTCTCACTCTGCCACCCAGGCTGGAGTGGCGG  60
      |||
AluSz6 1      TTT-----CTTTTTTGAGACAGCGTCTTGCTCTGTCCCCGGGCTGGAGTACAG  49

AluSx  61      TGGCACCATCTCGGCTCACTATAACCTCCGCCTCCTGAGTTCAAGTGATTCTTGTGCCTC  120
      ||| | |||| | |||| | |||| | ||| ||| ||| ||| ||| ||| ||| ||| |||
AluSz6 50      TGGTGCATCTTGACTCAATGCAACCTCTGTTACCCAAGTTCAAGTGATTCTCATGCATA  109

AluSx  121     AGCCTCTTGAGTAGCTGGGATTACAGGCGTGCCTCACCACGCCAGCTAATTTTTATATT  180
      ||| | |||| | |||| | |||| | ||| ||| ||| ||| ||| ||| ||| |||
AluSz6 110     AGCCACCAAATAGCTGGGACTACAGGCACACCT-ACCGCACCTGGCTTATTTTGTATT  168

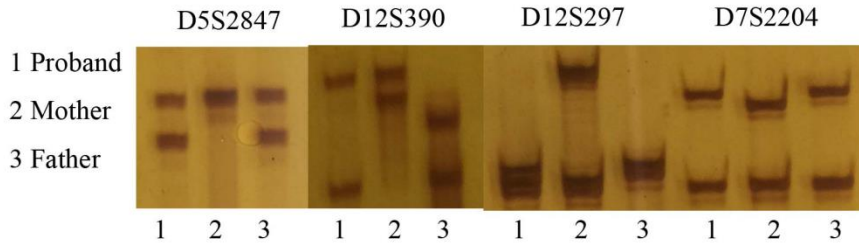
AluSx  181     TTTAGTAGAGACAGGGTTTCCACCAGGTTGGCCAGGCTAGTCTTGAACCTCCTGACCTCAGG  240
      ||| |||| |||| |||| |||| |||| |||| |||| |||| |||| |||| ||||
AluSz6 169     GTTAGTAGAGACAGGGTTTTGCCACATTAGCCAGGCTGGTATCAAATTCCTGACCTCAAG  228

AluSx  241     CGATCTGCCCTCCTCCACCTCCCAAAGTGTGGGATTACAGGCGTGAGCCACCGCGCCCA  300
      ||| | |||| |||| |||| |||| |||| |||| |||| |||| |||| ||||
AluSz6 229     TGATCCACCTGCCTCGGCCTCCCAATGTGCTGGAATTACAGGCATGAGCCACTGCACCTG  288

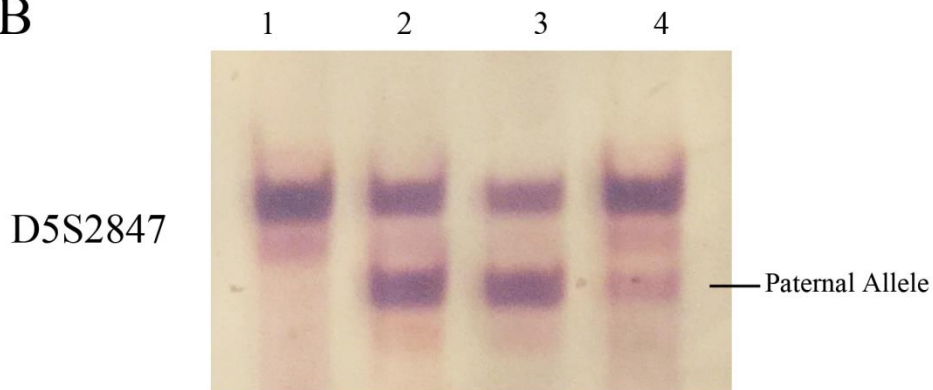
AluSx  301     GCC  303
      ||
AluSz6 289     ACC  291
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Figure 4. Alignment showed a high sequence identity up to 72% between two Alu repetitive elements.

A



B



1. Mother; 2. Father; 3. Proband

4. Mixed DNA with 10% from the proband and 90% from the mother

Figure 5. Selected microsatellite markers indicated maternal mosaicism was not caused by DNA contamination. We did not find any of the proband's paternal microsatellite markers mixed with the mother's (A). The positive control was generated by mixing 10% proband's sample with 90% his mother's sample (B).