**Materials and methods (extended)**

Subjects/probes

DNA of our patient and her parents was extracted from peripheral blood using standard procedures (1) (Quantification: NanoDrop TM ND-1000 (Thermo Fisher Scientific).

Informed consent for publication was obtained from the parents.

Array comparative genomic hybridization (Array CGH)

Array CGH analysis was performed with a 60k array and a 1M array for further specification of the duplication size (Agilent SurePrint G3 CGH/CGH+SNP 8x60K microarray kit and Agilent SurePrint G3 CGH/CGH+SNP 1x1M, Agilent Technologies, Santa Clara, CA, USA) following the instructions of the manufacturer. Commercially available megapool reference DNA (female) was used (Kreatech Leica Biosystems). Slides were scanned with an Agilent DNA Microarray Scanner G2505C and deletions/duplications were recorded with Agilent CytoGenomics software v5.0.0.38 (Agilent Technologies). Assessment of pathogenicity was performed by comparison of the data to Decipher database (2), HGMD (3) and review of the literature in PubMed.

Quantitative real time PCR (qRT-PCR)

qRT-PCR was performed using the ΔΔCT method.

Lyophilised primer were diluted to 100μM.

Patient DNA was used at a concentration of 100-200ng/μl.

For each pair of primer 10ng of DNA was used.

Fast SYBR Green Master Mix (Applied Biosystems/Thermo Fisher Scientific) was added following instructions.

7500 Fast Real-Time PCR System and 7500 Software v2.0.6 (Applied Biosystems/Thermo Fisher Scientific) were used following user manual.

Used primer:

Primer 1:

forward CCAGCTCAAAGGGATTGTG

reverse GTCGCTGTTTTCGTCCTTG

Primer 2:

forward GCGAGGGAGTCCAACAGC

reverse GCTGAACACCCCGAGGAC

Primer 3:

forward TGAGGTCTATTTGGTTTCAGAATTG

reverse GCTCACTGCATTGATGTTGG

Primer 5:

forward TGCCATGCTTTAGGAGGTG

reverse AGTGGCCTCCAGTGCATAAC

Primer 6:

forward AAACTCAGATTCAGTGGCACTTC

reverse CCCAAGGTCCTGAGTGAGAC

Primer 7:

forward TGTCTCTCTTTACTTAGCCACCAG

reverse AGCACAGCTGGAAGTTGAATC

Primer 8:

forward CCTTCAGGATAACAGCCCTTC

reverse ACCTTACCTCCAGGGCAGAG

Primer 10:

forward GAAGTCCCACTGTGTGTATAGGG

reverse CCCAAATGAACCTCACCTTTC

Primer 12:

forward TCCTATTCGGCCGTCTTTC

reverse GGACTTCAGCTAGATTTATTCCTACG

primer for control of PCR:

Primer 4fko:

forward CCTTTTAGGCCACCACCAC

reverse GATTTTGAGAAATGCCGTGTG

Primer NF2:

forward GGGAGAACAGCACATGATCC

reverse CTCCTCCTCGGTGATCTGG

Long range PCR

Long range PCR was performed with PrimeSTAR GXL DNA Polymerase (TaKaRa). As tandem duplication was expected, primers were designed near the expected breakpoints of the duplicated region to amplify the sequence lying between the duplicated segments (Figure 1). Primers were designed using Primer3plus (<http://www.bioinformatics.nl/cgi-bin/primer3plus/primer3plus.cgi/>):

Forward primer: TTAACGATCTCACTTCCCTAGTCAG

Reverse primer: CTAAAGCATGGCAAATCTATTCC

Chromosomal analysis

Chromosomal analysis from lymphocytes was performed using Giemsa staining with a resolution of 450-550 bands in the parents.

*Literature Material and Methods (extended)*

(1) [Miller SA](https://www.ncbi.nlm.nih.gov/pubmed/?term=Miller%20SA%5BAuthor%5D&cauthor=true&cauthor_uid=3344216), [Dykes DD](https://www.ncbi.nlm.nih.gov/pubmed/?term=Dykes%20DD%5BAuthor%5D&cauthor=true&cauthor_uid=3344216), [Polesky HF](https://www.ncbi.nlm.nih.gov/pubmed/?term=Polesky%20HF%5BAuthor%5D&cauthor=true&cauthor_uid=3344216). A simple salting out procedure for extracting DNA from human nucleated cells. Nucleic Acids Research 1988;16(3):1215.

(2) DECIPHER: Database of Chromosomal Imbalance and Phenotype in Humans using Ensembl Resources. Firth, H.V. et al (2009). Am.J.Hum.Genet 84, 524-533 (DOI: dx.doi.org/10/1016/j.ajhg.2009.03.010)

(3) Stenson et al (2003), The Human Gene Mutation Database (HGMD®): 2003 Update. *Hum Mutat* (2003) 21:577-581