Supplemental Table 1a. Primers of relevant STS markers, fluorescent dyes used, PCR product sizes and chromosome 9 location. Numbering according to NCBI Map Viewer Build 36.3.

STS marker	Primers	Product size	Chromosome localization
D9S196 PET	F - GGGATTACACCTCAAAACCA R - ACCACACTGCGGGACTT	254-260	95513904-95514161
RH109 FAM	F - CTGACCTGCTGTGCACCTTA R - TTTGGTTAGGGAGTGCCAAG	100	96405318-96405417
GDB583921 PET	F - CTGATGGGTGGATGGGTTTTC R - GCAGTCACCCCTCCTCCAGA	169	96418913-96419089
SHGC-110746 NED	F - ATGAGGGTGCAAATCAAAGAGAG R - GCTGAAATGGCATAGACCCAATA	156	97033833-97033988
U57800 VIC	F - TTCAATTGTCTGTTTCTCCC R - GGGTCAATCCATCAGGAAGA	102	97082319-97082420
203WH8 FAM	F - AGACAGTTTGAGGCTGCG R - AATTTTGCCCAGGAGACTTG	189	97113690-97113872
SHGC-8725 PET	F - CGCATAGCGTGTTTGAAAGTT R - GTATTTGTGCATTGGGCTCC	229	97247982-97248210
SHGC-156297 VIC	F - TTTTCTCGTTATGCTTTGGTGGT R - TGTGAGGCCGAAAAAGTACAAAT	334	97295823-97296156
RH92918 NED	F - GGCAGAGGCGTTTTTATTTG R - AAGGGGTGTACAAATGAACCC	146	97312189-97312334
SHGC-149159 NED	F - GAGCACAGCACATTCTGTTTCC R - TGAAGGAATTTGTGTGGAATGTG	300	97389774-97390073
D9S287 NED	F - AGGATGCTCCTCACGC R - ACCACTACATTGTTCAAGGG	168-180	97505937-97506112
G67556 NED	F - CAAAGGGATGAACCTTTGGA R - AGTGTTGGGGTGGATGTCTC	246	98100444-98100689
AFMA086YF1 VIC	F - GGAGAGTTGCTTGACCTCAG	219	98242351-

	R - TTAAGAAGTGAACACCAATAGGC		98242569
D9S180 VIC	F - CAGTGGTTTGGAATCGAACC R - AGCTATTTTTGGGGGCTGAG	220-230	99689308- 99689527
SHGC-4650 FAM	F - CCCTGAGCAAGATACTTAATCTCC R - CCTACCTGTTGGTTATATAGATTTTC	173	100541587- 100541759
SHGC-144486 VIC	F - AAATGTGCAGCCACAAGAAAAAT R - ATGCACAGTCTGGAACCCATAGT	315	100641193- 100641507
RH123392 NED	F - GCTTGACAAAGGCTCACCTAAGA R - CAACTTCCTCAGCAACGCTATTT	280	101540994- 101541273
SHGC-83931 NED	F - TTCTTCTCAGCCAAGAATGAACC R - AAAATTGAAGCTTGCAGTTGACC	315	101607994- 101608308
D9S127 FAM	F - AGATTGATTGATACAAGGATTTG R - CCCTCAAAATTGCTGTCTAT	149	105595935- 105596091

Supplemental Table 1b. Primers for additional markers used for fine mapping of deletion ends, fluorescent dyes used, PCR product sizes and chromosome 9 location. Numbering according to NCBI Map Viewer Build 36.3.

Marker	Primers	Product size	Chromosome localization
FANCC – exon 3 FAM	F - ATGTTATATTCAGGGATACTTG R - TAACAGTGAAGGGTATGTTTG	346	97042613-97042976
FANCC – exon 2 FAM	F - CCCATTTAAGGATGAAGT R - CATACATGGACAACAGTATAG	466	97049363-97049829
FANCC – exon 1 FAM	F - AGAGCCTTTTAGAAATGCTTC R - CCTGAAGTCAGAAAATAATTTC	329	97051186-97051514
BCC5-2 VIC	F - ATTCTCCCTGAAGGCCAAGT R - ACTCATTCCCCAACTTGCTG	228	101558357-101558584
BCC5-3 VIC	F - AAGGGCAGGAAGGAGGAATA R - GTGGCATATTTTCCCACACC	186	101566227-101566412
BCC5 NED	F - GAAGATGCCAAACCTCCAAA R - GTCAGTGCCTGCAGATTTGA	173	101574776-101574948

Supplemental Table 2. Typing of several polymorphic STS markers in our patient (IIIa) and her nephew (IVa). Markers D9S287 and D9S180 are deleted in the patient, but normal in nephew.

STS marker	patient (IIIa)	nephew (IVa)
D9S196	2, 2	1, 2
D9S287	1, del	1, 1
D9S180	1, del	1, 6
D9S127	1,1	1, 2

Supplemental Table 3. All known 23 OMIM genes mapped in 4.5 Mb region of chromosome 9 (q22.32-q22.33), which are deleted in our patient, are listed in table. Beside each gene name, description, OMIM number, there are also listed some functional properties of their protein products. Some of their main functional properties may exert dysfunction through phenotype of the patient (like *TGFBR1*, *CTSL2*, *FOXE1*, *XPA*).

Gene	Description	OMIM #	Function/dysfunction of gene product
FANCC	Fanconi anemia, complementation group C	227645	part of a nuclear protein complex together with FANCA, FANCG and FANCF; essential for protection against chromosome breakage [29, 30]
PTCH1	patched homolog 1 (Drosophila)	109400	receptor for hedgehog proteins, part of the hedgehog signaling pathway; mutations associated with Gorlin syndrome, basal cell carcinoma, medulloblastoma, and holoprosencephaly [1, 2, 7, 9]
HSD17B3	hydroxysteroid (17-beta) dehydrogenase 3	264300	catalyzes the conversion of androstenedione to testosterone; deficiency can result in male pseudohermaphroditism with gynecomastia [31]
SLC35D2	solute carrier family 35,	609182	nucleotide sugar transporter,

	member D2		translocation of nucleotide sugars from the cytosol into the lumen compartment [32]
ZNF367	zinc finger protein 367	610160	potential transcriptional activator of erythroid genes [33]
CDC14B	CDC14 cell division cycle 14 homolog B (S. cerevisiae)	603505	dual-specificity phosphatase, controls spindle assembly and disassembly during mitosis [34, 35]
CTSL2	cathepsin L2	603308	lysosomal cysteine proteinase, role in the maintenance of corneal avascularity [36]
TDRD7	tudor domain containing 7	611258	scaffold protein, specific function is unknown [37]
TMOD1	tropomodulin 1	190930	actin-capping protein, interacts with tropomyosin (TM) at the pointed end of actin filaments [38]
NCBP1	nuclear cap binding protein subunit 1, 80kDa	600469	component of the nuclear cap-binding protein complex (CBC); CBC promotes pre-mRNA splicing, 3'-end processing, RNA nuclear export, and nonsense-mediated mRNA decay [39]
XPA	xeroderma pigmentosum, complementation group A	278700	zinc finger protein involved in DNA excision repair [40]

FOXE1	forkhead box E1 (thyroid transcription factor 2)	241850	thyroid transcription factor; plays a role in thyroid morphogenesis; mutations associated with congenital hypothyroidism and cleft palate with thyroid dysgenesis [41]
HEMGN	hemogen	610715	human hematopoiesis-specific gene; overexpressed in various tissues and tumors such as thyroid tumor, thymus tumor, non-Hodgkin's lymphoma and PBMCs of patients with leukemia [42]
NANS	N-acetylneuraminic acid synthase (sialic acid synthase)	605202	enzyme that functions in the biosynthetic pathways of sialic acids; catalyzes the condensation of PEP (phosphoenolpyruvate) and ManNAc-6P (N-acetylmannosamine 6-phosphate) to yield NeuNAc-9P (N-acetylneuramic 9-phosphate acid) [43]
TRIM14	tripartite motif-containing 14	606556	cell compartment specification [44]
CORO2A	coronin, actin binding protein, 2A	602159	intracellular signaling [45]
TBC1D2	TBC1 domain family, member 2	609871	possible role in regulation of cell differentiation and growth [46]
GABBR2	gamma-aminobutyric acid	188890	slow inhibition of neural activity

	(GABA) B receptor, 2		through interaction with guanine nucleotide-binding (G) proteins that activate potassium channels, inactivate voltage-dependent calcium channels, and modulate adenylyl cyclase activity [47]
GALNT12	UDP-N-acetyl-alpha-D-galactosamine:polypeptide N-acetylgalactosaminyltransferase 12 (GalNAc-T12)	610290	catalyzes the transfer of N-acetylgalactosamine (GalNAc) from UDP-GalNAc to a hydroxyl amino acid on a polypeptide acceptor in the initial step of mucin-type O-linked protein glycosylation, specific for digestive organs [48]
COL15A1	collagen, type XV, alpha 1	120325	present in most basement membrane zones, closely associated with large collagen fibrils; may be important in spatial and temporal recruitment od modulators in growth, development and pathological processes; lack of this collagen causes skeletal myophaty and cardiovascular defects in mice [49, 50]
TGFBR1	transforming growth factor, beta receptor I (activin A	190181	important member of TGF- signaling pathway, necessary for signal

	receptor type II-like kinase, 53kDa)		transduction, mutated in carcinomas of the kidney and bladder [51]
ALG2	asparagine-linked glycosylation 2 homolog (S. cerevisiae, alpha-1,3-mannosyltransferase)	607905	role in glycoprotein biosynthesis, transfers mannosyl residues from GDP-Man to Man1GlcNAc2-PP-dolichol, deficiency causes congenital disorder of glycosylation CDG-Ii [52]
SEC61B	Sec61 beta subunit	609214	subunit of the heteromeric SEC61 complex, which forms the core of the transmembrane channel for the translocation of proteins across the ER membrane [53]

29 Garcia-Higuera I, Kuang Y, Näf D, Wasik J, D'Andrea AD: Fanconi anemia proteins FANCA, FANCC, and FANCG/XRCC9 interact in a functional nuclear complex. Mol Cell Biol 1999;19(7):4866-4873.

30 Pace P, Johnson M, Tan WM, Mosedale G, Sng C, Hoatlin M, de Winter J, Joenje H, Gergely F, Patel KJ.: FANCE: the link between Fanconi anaemia complex assembly and activity. EMBO J 2002;21(13) 3414-3423.

31 Mains LM, Vakili B, Lacassie Y, Andersson S, Lindqvist A, Rock JA: 17beta-hydroxysteroid dehydrogenase 3 deficiency in a male pseudohermaphrodite. Fertil Steril 2008;89(1):228.e13-17.

32 Ishida N, Kuba T, Aoki K, Miyatake S, Kawakita M, Sanai Y: Identification and characterization of human Golgi nucleotide sugar transporter SLC35D2, a novel member of the SLC35 nucleotide sugar transporter family. Genomics 2005;85(1):106-116.

33 Asano H, Murate T, Naoe T, Saito H, Stamatoyannopoulos G: Molecular cloning and characterization of ZFF29: a protein containing a unique Cys2His2 zinc-finger motif. Biochem J 2004;384(Pt 3):647-653.

34 Rosso L, Marques AC, Weier M, Lambert N, Lambot MA, Vanderhaeghen P, Kaessmann H: Birth and rapid subcellular adaptation of a hominoid-specific CDC14 protein. PLoS Biol 2008;6(6):e140.

35 Berdougo E, Nachury MV, Jackson PK, Jallepalli PV: The nucleolar phosphatase Cdc14B is dispensable for chromosome segregation and mitotic exit in human cells. Cell Cycle 2008;7(9):1184-1190.

36 Bernard D, Méhul B, Thomas-Collignon A, Simonetti L, Remy V, Bernard MA, Schmidt R: Analysis of proteins with caseinolytic activity in a human stratum corneum

extract revealed a yet unidentified cysteine protease and identified the so-called "stratum corneum thiol protease" as cathepsin l2. J Invest Dermatol 2003;120(4):592-600.

37 Skorokhod O, Nemazanyy I, Breus O, Filonenko V, Panasyuk G: Generation and characterization of monoclonal antibodies to TDRD7 protein. Hybridoma (Larchmt) 2008;27(3):211-216.

38 Kong KY, Kedes L: Leucine 135 of tropomodulin-1 regulates its association with tropomyosin, its cellular localization, and the integrity of sarcomeres. J Biol Chem 2006;281(14):9589-9599.

39 Ishigaki Y, Li X, Serin G, Maquat LE: Evidence for a pioneer round of mRNA translation: mRNAs subject to nonsense-mediated decay in mammalian cells are bound by CBP80 and CBP20. Cell 2001;106(5):607-617.

40 Vasquez KM, Christensen J, Li L, Finch RA, Glazer PM: Human XPA and RPA DNA repair proteins participate in specific recognition of triplex-induced helical distortions. Proc Natl Acad Sci U S A 2002;99(9):5848-5453.

41 Al Taji E, Biebermann H, Límanová Z, Hníková O, Zikmund J, Dame C, Grüters A, Lebl J, Krude H: Screening for mutations in transcription factors in a Czech cohort of 170 patients with congenital and early-onset hypothyroidism: identification of a novel PAX8 mutation in dominantly inherited early-onset non-autoimmune hypothyroidism. Eur J Endocrinol 2007;156(5):521-529.

42 Li CY, Zhan YQ, Xu CW, Xu WX, Wang SY, Lv J, Zhou Y, Yue PB, Chen B, Yang XM: EDAG regulates the proliferation and differentiation of hematopoietic cells and resists cell apoptosis through the activation of nuclear factor-kappa B. Cell Death Differ 2004;11(12):1299-1308.

43 Hamada T, Ito Y, Abe T, Hayashi F, Güntert P, Inoue M, Kigawa T, Terada T, Shirouzu M, Yoshida M, Tanaka A, Sugano S, Yokoyama S, Hirota H: Solution structure of the antifreeze-like domain of human sialic acid synthase. Protein Sci 2006;15(5):1010-1016.

44 Reymond A, Meroni G, Fantozzi A, Merla G, Cairo S, Luzi L, Riganelli D, Zanaria E, Messali S, Cainarca S, Guffanti A, Minucci S, Pelicci PG, Ballabio A: The tripartite motif family identifies cell compartments. EMBO J 2001;20(9):2140-2151.

45 Zaphiropoulos PG, Toftgård R: cDNA cloning of a novel WD repeat protein mapping to the 9q22.3 chromosomal region. DNA Cell Biol 1996;15(12):1049-1056.

46 Zhou Y, Toth M, Hamman MS, Monahan SJ, Lodge PA, Boynton AL, Salgaller ML: Serological cloning of PARIS-1: a new TBC domain-containing, immunogenic tumor antigen from a prostate cancer cell line. Biochem Biophys Res Commun 2002;290(2):830-838.

47 Martin SC, Steiger JL, Gravielle MC, Lyons HR, Russek SJ, Farb DH: Differential expression of gamma-aminobutyric acid type B receptor subunit mRNAs in the developing nervous system and receptor coupling to adenylyl cyclase in embryonic neurons. J Comp Neurol 2004;473(1):16-29.

48 Guo JM, Zhang Y, Cheng L, Iwasaki H, Wang H, Kubota T, Tachibana K, Narimatsu H: Molecular cloning and characterization of a novel member of the UDP-GalNAc:polypeptide N-acetylgalactosaminyltransferase family, pp-GalNAc-T12. FEBS Lett 2002;524(1-3):211-218.

49 Myers JC, Amenta PS, Dion AS, Sciancalepore JP, Nagaswami C, Weisel JW, Yurchenco PD: The molecular structure of human tissue type XV presents a unique conformation among the collagens. Biochem J 2007;404(3):535-544.

50 Eklund L, Piuhola J, Komulainen J, Sormunen R, Ongvarrasopone C, Fässler R, Muona A, Ilves M, Ruskoaho H, Takala TE, Pihlajaniemi T: Lack of type XV collagen causes a skeletal myopathy and cardiovascular defects in mice. Proc Natl Acad Sci U S A 2001;98(3):1194-1199.

51 Chen T, Jackson C, Costello B, Singer N, Colligan B, Douglass L, Pemberton J, Deddens J, Graff JR, Carter JH: An intronic variant of the TGFBR1 gene is associated with carcinomas of the kidney and bladder. Int J Cancer 2004;112(3):420-425.

52 Thiel C, Schwarz M, Peng J, Grzmil M, Hasilik M, Braulke T, Kohlschütter A, von Figura K, Lehle L, Körner C: A new type of congenital disorders of glycosylation (CDG-Ii) provides new insights into the early steps of dolichol-linked oligosaccharide biosynthesis. J Biol Chem 2003;278(25):22498-2505.

53 Greenfield JJ, High S: The Sec61 complex is located in both the ER and the ER-Golgi intermediate compartment. J Cell Sci 1999;112(Pt 10):1477-1486.