Down syndrome-like acute megakaryoblastic leukemia in a patient with Cornelia de Lange syndrome

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Supplementary table S1: List of SNVs and Indels identified by WES (whole exome sequencing) or targeted sequencing at D15 and acute megakaryoblastic leukemia (AMKL) stages

Relevant SNVs (Small Nucleotide Variation), Indels (Insertion/deletion) were called by eliminating the following entries: synonymous variants, inherited variants, variants with allele frequency > 1% in ExAC, ESP6500 or our in-house database unless they were registered as pathogenic in COSMIC, ClinVar, or HGMD. Mean depth of exome sequencing; patient: 97X, father: 80X, mother: 94X.

					D15 sample (WES) Mean depth: 97X		D15 sample (Targeted sequencing)		AMKL sample (Targeted sequencing)	
Gene	**RefSeq**	**Mutation type**	**Nucleotide change**	**Amino Acid Change**	**Sequencing depth**	**VAF**	**Sequencing depth**	**VAF**	**Sequencing depth**	**VAF**
NIPBL	NM_015384.4	splice	c.6344-2A>G	p.?	167X	0,31	47X	0,28	72X	0,44
GATA1	NM_002049.3	frameshift	c.44dupT	p.Gln17Profs*23	52X	0,38	51X	0,53	52X	0,84
EZH2	NM_004456.4	deletion	c.46_48delCGG	p.Arg16del	150X	0	142X	0	107X	0,64
NRAS	NM_002524.4	missense	c.436G>A	p.Ala146Thr	148X	0	105X	0	127X	0,37
JAK1	NM_002227.2	missense	c.2879C>G	p.Pro960Arg	68X	0	139X	0	132X	0,3

Supplementary table S2: List of CNVs and LOH (loss of heterozygosity) identified by CGH + SNP array at D15 and AMKL stages

Relevant CNVs (Copy Number Variation) were called by eliminating the CNVs overlapping more than 80% with regions seen at least 6 times in Benign consensus regions from ISCA CNV Atlas (bISCAr4) or the consensus region of the core DGV set (cDGVc7) or our in-house database unless they were registered as pathogenic in COSMIC, ClinVar, or HGMD.

D15 Sample						
Type of alteration	Chromosome	Start	Stop	Size (bp)	Cytoband	Log ratio
LOH	chr2	16815759	22832449	6016691	p24.2-p24.1	-
LOH	chr2	39863386	42432323	2568938	p22.1-p21	-
LOH	chr3	62808969	65639369	2830401	p14.2-p14.1	-
LOH	chr3	73004580	75257002	2252423	p13-p12.3	-
Gain	chr21	14671461	48090317	33418857	q11.2-q22.3	0.290654

AMKL Sample						
Type of alteration	Chromosome	Start	Stop	Size (bp)	Cytoband	Log ratio
LOH	chr1	71044680	75229396	4184717	p31.1	-
Duplication	chr1	160740134	249212668	88472535	q23.3-q44	0.443654
LOH	chr2	16815759	22832449	6016691	p24.2-p24.1	-
LOH	chr2	39863386	42981239	3117854	p22.1-p21	-
LOH	chr3	62808969	65639369	2830401	p14.2-p14.1	-
LOH	chr3	69627921	75257002	5629082	p14.1-p12.3	-
LOH	chr4	48354057	48907122	553066	p11	-
LOH	chr4	52780048	56778197	3998150	q12	-
Duplication	chr6	163083	46188827	46025745	p25.3-p21.1	0.419427
LOH	chr7	98400599	158885928	60485330	q22.1-q36.3	-
Deletion	chr7	100186998	159125464	58938467	q22.1-q36.3	-0.615681
Duplication	chr8	191530	146274835	43516763	p23.3-q24.3	0.461938
Duplication	chr9	204193	15680753	15476561	p24.3-p22.3	0.403025
LOH	chr11	4659187	6658270	1999084	p15.4	-
LOH	chr11	44081150	51505725	7424576	p11.2-p11.12	-
LOH	chr11	55196818	57448032	2251215	q11-q12.1	-
Duplication	chr19	27984957	59095418	31110462	q11-q13.43	0.390872
Duplication	chr21	14458104	48090317	33632214	q11.2-q22.3	0.450052