

Supplementary Table 1. Primers sequences for all exons and intron boundaries of *ELANE* and *HAX1* genes

Primer Name	Forward primer (5'->3')	Reverse primer (5'->3')
ELANE-EX1	TCTCCCCCTTTTCATCAACG	GTTCGCACCCCTGTTGTTC
ELANE-EX2,3	GAACAACAGGGGTGCGAAC	CTCATTTAACCCCTCAACGGC
ELANE-EX4,5	GGATCCCAGAACCACAGTG	CCCGACCTACTGACCATTTTC
HAX1-EX1	GATTTAGGAGGAAGGCGGTC	CCTCTTAATGACCCGCCTTG
HAX1-EX2,3	AGACCCCTTGCTCTTGTC	CTCACAAGCTCTCACTTCAGGAC
HAX1-EX4,5	GAGCTCGGGAGTAGTTTGAG	CCCACCATATTCCGAATGTTAC
HAX1-EX6,7	GTAACATTCGGAATATGGTGGG	GATAATTCGGAAGTGTTGGATG

Supplementary Table 2. Primers sequences for exons 2 and 23 of *HYOU1* and exon 2 of *SHOC2* gene

Primer name	Forward primer (5'->3')	Reverse primer (5'->3')
HYOU1-EX2	TGATCTGCCCTGGGTAAGAATGA	GCCACCTTCATGGACTCACTG
HYOU1-EX23	AGTCACTCCAGAGTAGTAGGATGTA	GGTAAGACATAGCCTCAGGAAGG
SHOC2-EX2	GATGTTACTCCATGCTGATTACTTC	CTTCTTCCATCTTTGGCATCTTTC

Supplementary Table 3. Variants related to immunodeficiency disorders in the CN05 family

Gene (Accession number)	Variant change	Zygoty	Inheritance Pattern	Clinical Phenotype	ACMG classification
<i>UNC13D</i> NM_199242.2	c.1728-13C>T c.569+10C>T	Het *	AR***	Hemophagocytic lymphohistiocytosis, familial, 3	VUS\$
<i>SPEN</i> NM_015001.2	c.1910G>A (p.Arg637Gln)	Het *	-	Megakaryoblastic leukemia, acute	VUS\$
<i>ARHGEF1</i> NM_199002.1	c.1636C>T (p.Arg546Cys)	Het *	AR***	Immunodeficiency 62	VUS\$
<i>IRGM</i> NM_001145805.1	c.313C>T (p.Leu105Leu)	Het *	-	Inflammatory bowel disease (Crohn's disease) 19	Pathogenic
<i>C8A</i> NM_000562.2	c.1246G>A (p.Val416Met)	Het *	AR***	C8 deficiency, type I	VUS\$
<i>FANCA</i> NM_000135.2	c.4232C>T (p.Pro1411Leu)	Het *	AR***	Fanconi anemia, complementation group A	VUS\$
<i>UPBI</i> NM_016327.2	c.105-2A>G	Het *	AR***	Beta-ureidopropionase deficiency	Pathogenic

*Het: Heterozygous; AD**: Autosomal dominant, AR***: Autosomal recessive, VUS\$: Variant of unknown significance

Supplementary Table 4. Variants related to immunodeficiency disorders in the CN06 family

Gene (Accession number)	Variant change	Zygoty	Inheritance Pattern	Clinical Phenotype	ACMG classification
<i>C9</i> NM_001737.3	c.701delA (p.Asn234fs)	Het *	AR***	C9 deficiency, Susceptibility to Macular degeneration, age-related, 15	Pathogenic
	c.697_698 insTTTTATG (p.Ser233fs)	Het *			Pathogenic
<i>IRF2BP2</i> NM_182972.2	c.1315C>T (p.His439Tyr)	Het *	AD**	Immunodeficiency, common variable, 14	Likely pathogenic
<i>CD36</i> NM_001001547	c.220C>T (p.Gln74*)	Het *	AR***	Platelet glycoprotein IV deficiency	Likely pathogenic
	c.660_669 delCATAAGTAAA (p.Asn220fs)	Het *			Pathogenic
<i>ANAPC1</i> NM_022662.3	c.1247C>T (p.Thr416Met)	Het *	AR***	Rothmund-Thomson syndrome, type 1	VUS [§]
	c.1084T>G (p.Ser362Ala)	Het *			VUS [§]
<i>LRRC56</i> NM_198075.3	c.371C>T (p.Ala124Val)	Het *	AR***	Ciliary dyskinesia, primary, 39	VUS [§]
	c.655G>A (p.Val219Met)	Het *			Likely pathogenic
<i>LPP</i> NM_005578.3	c.1484A>G (p.Tyr495Cys)	Het *	AD**	Leukemia, acute myeloid	Likely pathogenic
<i>NHP2</i> NM_017838.3	c.3G>A (p.Met1Ile)	Het *	AR***	Dyskeratosis congenita, 2	Likely pathogenic

Het: Heterozygous; AD: Autosomal dominant, AR***: Autosomal recessive, VUS[§]: Variant of unknown significance