Table 1 Congenital defects of phagocyte number, function, or both

	Disease	Affected cells	Affected function	Associated features	Inheritance	Genetic defects/presumed pathogenesis	OMIM	HGNC symbol	HGNC ID(s)	ENSEMBL ID
1.-3.	**Severe congenital neutropenias**	N	Myeloid differentiation	Subgroup with myelodysplasia	AD	*ELA2*: mistrafficking of elastase	202700, 130130	*ELANE*	3309	ENSG00000197561
		N	Myeloid differentiation	B/T lymphopenia	AD	*GFI1*: repression of elastase	613107, 600871	*GFI1*	4237	ENSG00000162676
4.	**Kostmann syndrome**	N	Myeloid differentiation		AR	HAX1: control of apoptosis	610738, 605998	*HAX1*	16915	ENSG00000143575
5.	**Cyclic neutropenia**	N		Oscillations of other leukocytes and platelets	AD	*ELA2*: mistrafficking of elastase	162800	*ELANE*	3309	ENSG00000197561
6.	**X-linked neutropenia /myelodysplasia**	N + M		Monocytopenia	XL	*WASP:* Regulator of actin cytoskeleton (loss of autoinhibition)	300299, 300392	*WAS*	12731	ENSG00000015285
	SCN4(G6PC3 deficiency)	N+ F	Myeloid differentiation, chemotaxis, O_2^- production	Structural heart defects, urogenital abnormalities, and venous angiectasis of trunks and limbs	AR	*G6PC3*: abolished enzymatic activity of glucose-6-phosphatase, aberrant glycosylation, and enhanced apoptosis of neutrophils and fibroblasts	612541, 611045	*G6PC3*	24861	ENSG00000141349
	e) Glycogen storage disease type1b	N+ M	Myeloid differentiation, chemotaxis, O_2^- production	Fasting hypoglycemia, lactic acidosis, hyperlipidemia, hepatomegaly	AR	*G6PT* 1: glucose-6-phosphate transporter 1	232220, 602671	*SLC37A4*	4061	ENSG00000137700
7.	**P14 deficiency**	N + L + melanocytes	Endosome biogenesis	Neutropenia, Hypogammaglobulinemia, ↓CD8 cytotoxicity, Partial albinism, Growth failure	AR	*MAPBPIP:* Endosomal adaptor protein 14	610389	*LAMTOR2*	29796	ENSG00000116586
	(i) Barth syndrome	N	Myeloid differentiation	Cardiomyopathy, growth retardation	XL	Tafazzin(*TAZ*): abnormal lipid structure of mitochondrial membrane	302060, 300394	*TAZ*	11577	ENSG00000102125
	(j) Cohen syndrome	N	Myeloid differentiation	Retinopathy, developmental delay, facial dysmorphisms	AR	*COH*1: Pathogenesis unknown	216550, 607817	*VPS13B*	2183	ENSG00000132549
	(k) Poikiloderma with neutropenia	N	Myeloid differentiation, O_2^- production	Poikiloderma, MDS	AR	C16*orf* 57: Pathogenesis unknown	604173, 613276	*C16orf57*	25792	ENSG00000103005
8.	**Leukocyte adhesion deficiency type 1**	N + M L + NK	Adherence Chemotaxis Endocytosis T/NK cytotoxicity	Delayed cord separation, Skin ulcers, Periodontitis, Leukocytosis	AR	*ITGB2:* Adhesion protein CD18	116920, 600065	*ITGB2*	6155	ENSG00000160255
9.	**Leukocyte adhesion deficiency type 2 = Congenital disorder of glycosylation type IIc**	N + M L + NK	Rolling, Chemotaxis	Recurrent bacterial infections periodontitis, pneumonia, cellulites without pus formation, leukocytosis, hh-blood group, short stature, mental retardation	AR	*SLC35C1:* FUCT1 GDP-fucose transporter	266265, 605881	*SLC35C1*	20197	ENSG00000181830
10.	**Leukocyte adhesion deficiency type 3**	N + M + L + NK + platelets	Adherence	Infections, leukocytosis, bleeding tendency	AR	*RASGRB2*: Rap1- mediated activation of β1-3 integrins	612840, 607901	*FERMT3*	23151	ENSG00000149781
11.	**Rac 2 deficiency**	N	Adherence Chemotaxis O_2^- production	Poor wound healing, Leukocytosis	AD	*RAC2*: Regulation of actin cytoskeleton	602049	*RAC2*	9802	ENSG00000128340
12.	**β-Actin deficiency**	N + M	Motility	Mental retardation, Short stature	AD	*ACTB*: Cytoplasmic actin	102630	*ACTB*	132	ENSG00000075624
13.	**Localized juvenile periodontitis**	N	Formylpeptide-induced chemotaxis	Periodontitis only	AR	*FPR1*: Chemokine receptor	136537	*FPR1*	3826	ENSG00000171051
14.	**Papillon-Lefèvre syndrome**	N + M	Chemotaxis	Periodontitis, palmoplantar hyperkeratosis	AR	*CTSC*: Cathepsin C activation of serine proteases	245000, 602365	*CTSC*	2528	ENSG00000109861
15.	**Specific granule deficiency**	N	Chemotaxis	N with bilobed nuclei	AR	*C/EBPE*: myeloid Transcription factor	245480, 600749	*CEBPE*	1836	ENSG00000092067
16.	**Shwachman-Diamond syndrome**	N	Chemotaxis	Pancytopenia, exocrine pancreatic insufficiency, Chondrodysplasia	AR	*SBDS*	260400, 607444	*SBDS*	19440	ENSG00000126524
17.	**X-linked chronic granulomatous disease (CGD)**	N + M	Killing (faulty O_2^- production)	Subgroup: McLeod phenotype	XL	*CYBB*: Electron transport protein (gp91phox)	306400, 300481	*CYBB*	2578	ENSG00000165168
18.-20.	**Autosomal CGDs (3 forms)**	N + M	Killing (faulty O_2^- production)		AR	*CYBA*: Electron transport protein (p22phox)	233690, 608508	*CYBA*	2577	ENSG00000051523
						NCF1: Adapter protein (p47phox)	233700, 608512	*NCF1*	7660	ENSG00000158517
						NCF2: Activating protein (p67phox)	233710, 608515	*NCF2*	7661	ENSG00000116701
						NCF4: activatingprotein(p40 phox)	601488	*NCF4*	7662	ENSG00000100365
21.	**Neutrophil G-6PD deficiency**	N + M	Killing (faulty O_2^- production)	Hemolytic anemia	XL	*G-6PD*: NADPH generation	305900	*G6PD*	4057	ENSG00000160211
22.	**IL-12 and IL-23 receptor β1 chain deficiency**	L + NK	IFN-γ secretion	Susceptibility to *Mycobacteria* and *Salmonella*	AR	*IL-12Rβ1*: IL-12 and IL-23 receptor β1 chain	601604	*NCF4*	7662	ENSG00000100365
23.	**IL-12p40 deficiency**	M	IFN-γ secretion	Susceptibility to *Mycobacteria* and *Salmonella*	AR	*IL-12p40* subunit of IL12/IL23: IL12/IL23 production	161561	*IL12B*	5970	ENSG00000113302
24.	**IFN-γ receptor 1 deficiency**	M + L	IFN-γ binding and signalling	Susceptibility to *Mycobacteria* and *Salmonella*	AR, AD*	*IFNGR1*: IFN-γR binding chain	107470	*IFNGR1*	5439	ENSG00000027697
25.	**IFN-γ receptor 2 deficiency**	M + L	IFN-γ signaling	Susceptibility to *Mycobacteria* and *Salmonella*	AR	*IFNGR2*: IFN-γR signaling chain	*147*569	*IFNGR2*	5440	ENSG00000159128
26.	**STAT1 deficiency (2 forms)**	M + L	IFN α/β/γ signaling	Susceptibility to *Mycobacteria, Salmonella*, and viruses	AR	*STAT1*				
			IFN-γ signaling	Susceptibility to *Mycobacteria* and *Salmonella*	AD*	*STAT1*	614162, 600555	*STAT1*	11362	ENSG00000115415

N, Neutrophils; *AD*, autosomal dominant; *AR*, autosomal recessive inheritance; *M*, monocytes-macrophages; *XL*, X-linked inheritance; *L*, lymphocytes; *NK*, natural killer cells; *LAD*, leukocyte adhesion deficiency; *FUCT1*, fucose transporter 1; *GDP*, guanosine diphosphate; *SBDS*, Schwachman-Bodan-Diamond syndrome; *STAT1*, signal transducer and activator of transcription 1.

*The AD form of IFNGR1 deficiency or of STAT1 deficiency is caused by dominant negative mutations.

G-CSFR mutations are no longer in the list causing severe congenital neutropenia since mutations in G-CSFR are now regarded as aquired somatic mutations linked to G-CSF refractory neutropenia and cancer development (324)