

Supplementary Tables:

Table S1: Neutrophil and monocyte counts of individuals where data was available.

Readings outside the reference range (Ref) have been shown in bold.

Parameter		Ref	II-1	III-2	IV-1	IV-2	IV-3	IV-4	V-1
Neutrophils	$\times 10^9/L$	2.0-7.5	0.957	0.8	0.76	0.5	2.45	1.1	0.7
Monocytes	$\times 10^9/L$	0-1.0	1.122	2.1	1.68	1.6	1.72	1.3	1.5

Table S2: Genes known to be mutated in congenital neutropenia (Skokowa *et al.* 2017).

Autosomal Dominant Inheritance				
<i>ELANE</i>	<i>GF11</i>	<i>GATA2</i>	<i>TCIRG1</i>	<i>CXCR4</i>
<i>CSF3R</i>				
Autosomal Recessive Inheritance				
<i>HAX1</i>	<i>JAGN1</i>	<i>G6PC3</i>	<i>SLC37A4</i>	<i>SBDS</i>
<i>STK4</i>	<i>CLPB</i>	<i>AP3B1</i>	<i>LAMTOR2</i>	<i>USB1</i>
<i>VPS13B</i>	<i>VPS45</i>	<i>CXCR2</i>	<i>EIFAK3</i>	<i>LYST</i>
<i>RAB27A</i>	<i>AK2</i>	<i>RMRP</i>	<i>TCN2</i>	<i>CSF3R</i>
X-linked Inheritance				
<i>WAS</i>	<i>TAZ</i>	<i>CD40LG</i>		

Table S3: Genes known to be mutated in non-syndromic hearing loss (Shearer *et al.*, 2017).

Autosomal Dominant Inheritance				
<i>ACTG1</i>	<i>CCDC50</i>	<i>CD164</i>	<i>CEACAM16</i>	<i>COCH</i>
<i>COL11A2</i>	<i>GSDME</i>	<i>DIAPH1</i>	<i>DMXL2</i>	<i>DSPP</i>
<i>EYA4</i>	<i>GJB2</i>	<i>GJB3</i>	<i>GJB6</i>	<i>GRHL2</i>
<i>HOMER2</i>	<i>KCNQ4</i>	<i>MIR96</i>	<i>MCM2</i>	<i>MYH14</i>
<i>MYH9</i>	<i>MYO1A</i>	<i>MYO6</i>	<i>MYO7A</i>	<i>OSBPL2</i>
<i>P2RX2</i>	<i>POU4F3</i>	<i>SIX1</i>	<i>SLC17A8</i>	<i>TBC1D24</i>
<i>TECTA</i>	<i>TJP2</i>	<i>TMC1</i>	<i>WFS1</i>	
Autosomal Recessive Inheritance				
<i>ADCY1</i>	<i>BDP1</i>	<i>BSND</i>	<i>CABP2</i>	<i>CDC14A</i>
<i>CDH23 1</i>	<i>CIB2</i>	<i>CLDN14</i>	<i>CLIC5</i>	<i>COL11A2</i>
<i>DCDC2</i>	<i>PJK</i>	<i>ELMOD3</i>	<i>EPS8</i>	<i>EPS8L2</i>
<i>ESPN</i>	<i>ESRRB</i>	<i>GIPC3 2</i>	<i>GJB2 3</i>	<i>GJB6 3</i>
<i>GPSM2</i>	<i>GRXCR1</i>	<i>GRXCR2</i>	<i>HGF</i>	<i>ILDR1</i>
<i>KARS</i>	<i>LHFPL5</i>	<i>LOXHD1</i>	<i>LRTOMT</i>	<i>MARVELD2</i>
<i>MET</i>	<i>MSRB3</i>	<i>MYO15A</i>	<i>MYO3A</i>	<i>MYO6</i>
<i>MYO7A 5</i>	<i>NARS2 6</i>	<i>OTOG</i>	<i>OTOGL</i>	<i>OTOA</i>
<i>OTOF</i>	<i>PCDH15</i>	<i>PNPT1</i>	<i>PTPRQ</i>	<i>RDX</i>
<i>RIPOR2</i>	<i>ROR1 7</i>	<i>SIPR2</i>	<i>SERPINB6</i>	<i>SLC22A4</i>
<i>SLC26A4</i>	<i>SLC26A5</i>	<i>STRC</i>	<i>SYNE4</i>	<i>TECTA</i>
<i>TBC1D24</i>	<i>TMC1</i>	<i>TMEM132E</i>	<i>TMIE</i>	<i>TMPRSS3</i>
<i>TPRN</i>	<i>TRIOBP</i>	<i>TSPEAR</i>	<i>USH1C 12</i>	<i>WBP2</i>
<i>WHRN</i>				
X-linked Inheritance				
<i>PRPS1</i>	<i>POU3F4</i>	<i>SMPX</i>	<i>AIFM1</i>	<i>COL4A6</i>

Table S4: Computational predictions, variant annotation and classification.

HGNC Symbol	Position (hg19)	Amino Acid/ Nucleotide Change	CADD	GERP	Population Frequency (gnomAD)	ACMG Classification
GFI1	Chr1:92,941,710	p.Asn382Ser/ c.1145A>G NM_005263.5	23.9	4.18	0	Pathogenic
MYO6	Chr6:76,623,866	p.Ile1176Leu/ c.3526A>C NM_004999.4	25.3	5.89	3/277220	Likely pathogenic

Supplemental References:

Shearer, A.E., Hildebrand, M.S., Smith, R.J.H. Hereditary Hearing Loss and Deafness Overview. Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK1434/>

Skokowa, J., Dale, D. C., Touw, I. P., Zeidler, C. & Welte, K. (2017). Severe congenital neutropenias. *Nature Reviews Disease Primers* **3**: 17032.