

Table S2. Genes within a 954,849 bp heterozygous deletion at chr22:20,459,997–21,414,845 (GRCh37/hg19), with OMIM identifiers, modes of inheritance and associated phenotypes.

	Gene	OMIM gene	Inheritance	Phenotype
1	<i>ZNF74</i>	194548	//	
2	<i>THAP7</i>	609518	//	
3	<i>SNAP29</i>	604202	AR	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome
4	<i>SLC7A4</i>	603752	//	
5	<i>SERPIND1</i>	142360	AD	Thrombophilia 10 due to heparin cofactor II deficiency
6	<i>SCARF2</i>	613619	AR	Van den Ende-Gupta syndrome
7	<i>RIMBP3</i>	612699	//	
8	<i>PI4KA</i>	600286	AR	Gastrointestinal defects and immunodeficiency syndrome 2; Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis; Spastic paraplegia
9	<i>P2RX6</i>	608077	//	
10	<i>MED15</i>	607372	//	
11	<i>LZTR1</i>	600574	AD/AR	susceptibility to Schwannomatosis-2; Noonan syndrome; Noonan syndrome 2
12	<i>LRRC74B</i>	//	//	
13	<i>KLHL22</i>	618020	//	
14	<i>CRKL</i>	602007	//	CAKUT
15	<i>AIFM3</i>	617298	//	

Legend: AD, autosomal dominant; AR, autosomal recessive; CAKUT, congenital anomalies of the kidney and urinary tract; GRCh37/hg19, Genome Reference Consortium Human Genome Build 37 (human genome assembly hg19); OMIM, Online Mendelian Inheritance in Man; //, no established disease association or inheritance pattern currently reported in OMIM.