

The Growth failure in early childhood panel genes (PanelApp v1.4, March 2020)

Gene	Associated Phenotype(s)	OMIM
<i>BLM</i>	Bloom syndrome	210900
<i>BRCA2</i>	Fanconi anemia, complementation group D1 Please note: Only biallelic <i>BRCA2</i> variants will be reported.	605724
<i>BRIP1</i>	Fanconi anemia, complementation group J	609054
<i>ERCC4</i>	Fanconi anemia, complementation group Q	615272
<i>FANCA</i>	Fanconi anemia, complementation group A	227650
<i>FANCB</i>	Fanconi anemia, complementation group B	300514
<i>FANCC</i>	Fanconi anemia, complementation group C	227645
<i>FANCD2</i>	Fanconi anemia, complementation group D2	227646
<i>FANCE</i>	Fanconi anemia, complementation group E	600901
<i>FANCF</i>	Fanconi anemia, complementation group F	603467
<i>FANCG</i>	Fanconi anemia, complementation group G	614082
<i>FANCI</i>	Fanconi anemia, complementation group I	609053
<i>FANCL</i>	Fanconi anemia, complementation group L	614083
<i>NBN</i>	Nijmegen breakage syndrome	251260
<i>PALB2</i>	Fanconi anemia, complementation group N	610832
<i>SLX4</i>	Fanconi anemia, complementation group P	613951
<i>TOP3A</i>	Microcephaly, growth restriction, and increased sister chromatid exchange type 2, MGRISCE2 (Bloom-like syndrome)	618097
<i>UBE2T</i>	Fanconi anemia, complementation group T	616435
<i>CCDC8</i>	3-M syndrome type 3	614205
<i>CUL7</i>	3-M syndrome type 1	273750
<i>OBSL1</i>	3M syndrome type 2	612921
<i>SRCAP</i>	Floating-Harbor syndrome	136140
<i>TRIM37</i>	Mulibery Nanism	253250
<i>BRAF</i>	LEOPARD syndrome type 3 Cardiofaciocutaneous syndrome Noonan Syndrome type 7	613707 115150 613706
<i>CBL</i>	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	613563
<i>HRAS</i>	Costello syndrome	218040
<i>KRAS</i>	Cardiofaciocutaneous syndrome type 2 Noonan syndrome type 3	615278 609942
<i>LZTR1</i>	Noonan syndrome type 2 (autosomal recessive) Noonan syndrome type 10 (autosomal dominant)	605275 616564
<i>MAP2K1</i>	LEOPARD syndrome Cardiofaciocutaneous syndrome type 3	PMID: 20301303 615279
<i>MAP2K2</i>	Cardiofaciocutaneous syndrome type 4	615280
<i>NRAS</i>	Noonan syndrome type 6	613224
<i>PPP1CB</i>	Noonan syndrome-like disorder with loose anagen hair type 2	617506
<i>PTPN11</i>	LEOPARD syndrome type 1 Noonan syndrome type 1	151100 163950
<i>RAF1</i>	LEOPARD syndrome type 2 Noonan syndrome type 5	611554 611553
<i>RIT1</i>	Noonan syndrome type 8	615355
<i>SHOC2</i>	Noonan syndrome-like with loose anagen hair	607721
<i>SOS1</i>	Noonan syndrome type 4	610733
<i>SOS2</i>	Noonan syndrome type 9	616559
<i>HMGA2</i>	Silver-Russell syndrome	PMID:29655892

<i>IGF2</i>	Growth retardation with deafness and mental retardation due to IGF1 deficiency	608747
<i>PLAG1</i>	Silver-Russell syndrome	PMID: 20301499
<i>ACAN</i>	Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans Spondyloepimetaphyseal dysplasia, aggrecan type	165800 612813
<i>ANKRD11</i>	KBG syndrome	148050
<i>CDKN1C</i>	Intrauterine growth retardation, Metaphyseal dysplasia, Adrenal hypoplasia congenita, and Genital anomalies (IMAGE)	614732
<i>FGFR3</i>	Hypochondroplasia	146000
<i>IGF1</i>	Growth retardation with deafness and mental retardation due to IGF1 deficiency	608747
<i>IGF1R</i>	Resistance to Insulin-like growth factor type 1	270450
<i>PIK3R1</i>	SHORT syndrome	269880