Craniofacial microsomia

Gene panel

Gene panel information

Gene panel	**Craniofacial microsomia**
Version	2
Total genes	37
Activation date	Tuesday 16 june 2026
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
AMIGO2	99.96 %	615690	*No OMIM phenotypes*
CDT1	99.68 %	605525	Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive
CLTCL1	99.44 %	601273	*No OMIM phenotypes*
CRKL	99.64 %	602007	*No OMIM phenotypes*
DACH1	99.92 %	603803	*No OMIM phenotypes*
DACH2	99.73 %	300608	*No OMIM phenotypes*
EFTUD2	99.55 %	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
EYA3	97.53 %	601655	*No OMIM phenotypes*
FANCB	99.51 %	300515	Fanconi anemia, complementation group B, 300514 (3), X-linked recessive
FBLN2	99.66 %	135821	*No OMIM phenotypes*
FGF3	99.74 %	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive
FOXI3	99.8 %	612351	Craniofacial microsomia 2, 620444 (3), Autosomal recessive, Autosomal dominant
GBX2	99.83 %	601135	*No OMIM phenotypes*
GSC2	98.81 %	601845	*No OMIM phenotypes*
HIRA	99.48 %	600237	*No OMIM phenotypes*
HMX1	99.9 %	142992	Oculoauricular syndrome, 612109 (3), Autosomal recessive
HOXA2	100 %	604685	Microtia with or without hearing impairment (AD), 612290 (3), Autosomal recessive, Autosomal dominant; ?Microtia, hearing impairment, and cleft palate (AR), 612290 (3), Autosomal recessive, Autosomal dominant
HSPA9	99.8 %	600548	Even-plus syndrome, 616854 (3), Autosomal recessive; Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant
ITGB4	99.89 %	147557	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730 (3), Autosomal recessive; Epidermolysis bullosa, junctional 5A, intermediate, 619816 (3), Autosomal recessive
MAPK1	99.73 %	176948	Noonan syndrome 13, 619087 (3), Autosomal dominant
MARS1	99.43 %	156560	Spastic paraplegia 70, autosomal recessive, 620323 (3), Autosomal recessive; Interstitial lung and liver disease, 615486 (3), Autosomal recessive; ?Trichothiodystrophy 9, nonphotosensitive, 619692 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant
MYT1	99.94 %	600379	*No OMIM phenotypes*

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OTX2	99.98 %	600037	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant
PDE4DIP	67.36 %	608117	*No OMIM phenotypes*
POLR1B	99.59 %	602000	Treacher-Collins syndrome 4, 618939 (3), Autosomal dominant
POLR1C	100 %	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
POLR1D	99.99 %	613715	Treacher Collins syndrome 2, 613717 (3), Autosomal recessive, Autosomal dominant
SF3B2	99.75 %	605591	Craniofacial microsomia, 164210 (3), Autosomal dominant
SHROOM3	99.89 %	604570	*No OMIM phenotypes*
SIX1	99.98 %	601205	Deafness, autosomal dominant 23, 605192 (3), Autosomal dominant; Branchiootic syndrome 3, 608389 (3), Autosomal dominant
SIX5	99.24 %	600963	Branchiootorenal syndrome 2, 610896 (3)
TBX1	98.71 %	602054	Tetralogy of Fallot, 187500 (3), Autosomal dominant; DiGeorge syndrome, 188400 (3), Autosomal dominant; Conotruncal anomaly face syndrome, 217095 (3); Velocardiofacial syndrome, 192430 (3), Autosomal dominant
TCOF1	99.76 %	606847	Treacher Collins syndrome 1, 154500 (3), Autosomal dominant
VWA1	99.7 %	611901	Neuronopathy, distal hereditary motor, autosomal recessive 7, 619216 (3), Autosomal recessive
YPEL1	99.98 %	608082	*No OMIM phenotypes*
ZIC3	99.87 %	300265	Congenital heart defects, nonsyndromic, multiple types, 1, X-linked, 306955 (3), X-linked recessive; Heterotaxy, visceral, 1, X-linked, 306955 (3), X-linked recessive; VACTERL association, X-linked, 314390 (3), X-linked recessive
ZYG11B	97.89 %	618673	*No OMIM phenotypes*

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.