# Ectodermal dysplasia

Gene panel

## Gene panel information

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| **Gene panel** | **Ectodermal dysplasia** |
| **Version** | 5 |
| **Total genes** | 96 |
| **Activation date** | Tuesday 23 may 2023 |
| **Publisher** | Center for Medical Genetics, Ghent |

## Genes

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **ACP4** | 99.71 % | 606362 | Amelogenesis imperfecta, type IJ, 617297 (3), Autosomal recessive |
| **AMBN** | 99.93 % | 601259 | Amelogenesis imperfecta, type IF, 616270 (3), Autosomal recessive |
| **AMELX** | 99.94 % | 300391 | Amelogenesis imperfecta, type 1E, 301200 (3), X-linked dominant |
| **ANAPC1** | 72.56 % | 608473 | Rothmund-Thomson syndrome, type 1, 618625 (3), Autosomal recessive |
| **APCDD1** | 99.82 % | 607479 | Hypotrichosis 1, 605389 (3), Autosomal dominant |
| **ARID1A** | 99.61 % | 603024 | Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant |
| **ARID1B** | 99.8 % | 614556 | Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant |
| **ARID2** | 99.49 % | 609539 | Coffin-Siris syndrome 6, 617808 (3), Autosomal dominant |
| **AXIN2** | 99.8 % | 604025 | Colorectal cancer, somatic, 114500 (3); Oligodontia-colorectal cancer syndrome, 608615 (3), Autosomal dominant |
| **BICRA** | 99.71 % | 605690 | Coffin-Siris syndrome 12, 619325 (3), Autosomal dominant |
| **C3orf52** | 99.63 % | 611956 | Hypotrichosis 15, 620177 (3), Autosomal recessive |
| **CDH3** | 99.86 % | 114021 | Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive; Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive |
| **CDSN** | 99.91 % | 602593 | Hypotrichosis 2, 146520 (3), Autosomal dominant; Peeling skin syndrome 1, 270300 (3), Autosomal recessive |
| **CNNM4** | 99.81 % | 607805 | Jalili syndrome, 217080 (3), Autosomal recessive |
| **COL1A1** | 99.22 % | 120150 | Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Caffey disease, 114000 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 (3), Autosomal dominant; Osteogenesis imperfecta, type I, 166200 (3), Autosomal dominant; {Bone mineral density variation QTL, osteoporosis}, 166710 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant |
| **COL1A2** | 98.99 % | 120160 | Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; {Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 (3), Autosomal dominant; Ehlers-Danlos syndrome, cardiac valvular type, 225320 (3), Autosomal recessive; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant |
| **DLX3** | 99.28 % | 600525 | Trichodontoosseous syndrome, 190320 (3), Autosomal dominant; Amelogenesis imperfecta, type IV, 104510 (3), Autosomal dominant |
| **DPF2** | 99.89 % | 601671 | Coffin-Siris syndrome 7, 618027 (3), Autosomal dominant |
| **DSG4** | 99.88 % | 607892 | Hypotrichosis 6, 607903 (3), Autosomal recessive |

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|---|---|---|---|
| **DSPP** | 99.98 % | 125485 | Dentinogenesis imperfecta, Shields type III, 125500 (3), Autosomal dominant; Dentinogenesis imperfecta, Shields type II, 125490 (3), Autosomal dominant; Dentin dysplasia, type II, 125420 (3), Autosomal dominant; Deafness, autosomal dominant 39, with dentinogenesis, 605594 (3), Autosomal dominant |
| **EDA** | 99.51 % | 300451 | Tooth agenesis, selective, X-linked 1, 313500 (3), X-linked dominant; Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 (3), X-linked recessive |
| **EDAR** | 99.34 % | 604095 | [Hair morphology 1, hair thickness], 612630 (3); Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 (3), Autosomal dominant; Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 (3), Autosomal recessive |
| **EDARADD** | 99.95 % | 606603 | Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 (3), Autosomal recessive; Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 (3), Autosomal dominant |
| **ENAM** | 100 % | 606585 | Amelogenesis imperfecta, type IC, 204650 (3), Autosomal recessive; Amelogenesis imperfecta, type IB, 104500 (3), Autosomal dominant |
| **FAM20A** | 99.95 % | 611062 | Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 (3), Autosomal recessive |
| **FAM83H** | 99.99 % | 611927 | Amelogenesis imperfecta, type IIIA, 130900 (3), Autosomal dominant |
| **FGFR1** | 99.77 % | 136350 | Pfeiffer syndrome, 101600 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3) |
| **GJB6** | 100 % | 604418 | Ectodermal dysplasia 2, Clouston type, 129500 (3), Autosomal dominant; Deafness, autosomal dominant 3B, 612643 (3), Autosomal dominant; Deafness, autosomal recessive 1B, 612645 (3), Autosomal recessive; Deafness, digenic GJB2/GJB6, 220290 (3), Digenic dominant, Autosomal recessive |
| **GPR68** | 99.98 % | 601404 | Amelogenesis imperfecta, hypomaturation type, IIA6, 617217 (3), Autosomal recessive |
| **GREM2** | 100 % | 608832 | Tooth agenesis, selective, 9, 617275 (3), Autosomal dominant |
| **GRHL2** | 99.68 % | 608576 | Deafness, autosomal dominant 28, 608641 (3), Autosomal dominant; Ectodermal dysplasia/short stature syndrome, 616029 (3), Autosomal recessive; Corneal dystrophy, posterior polymorphous, 4, 618031 (3), Autosomal dominant |
| **HOXC13** | 99.63 % | 142976 | Ectodermal dysplasia 9, hair/nail type, 614931 (3), Autosomal recessive |
| **HR** | 99.88 % | 602302 | Atrichia with papular lesions, 209500 (3), Autosomal recessive; Alopecia universalis, 203655 (3), Autosomal recessive |
| **IKBKG** | 54.61 % | 300248 | Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive; Autoinflammatory disease, systemic, X-linked, 301081 (3), X-linked |
| **ITGB6** | 99.3 % | 147558 | Amelogenesis imperfecta, type IH, 616221 (3), Autosomal recessive |
| **JUP** | 99.59 % | 173325 | Naxos disease, 601214 (3), Autosomal recessive; ?Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant |
| **KLK4** | 99.86 % | 603767 | Amelogenesis imperfecta, type IIA1, 204700 (3), Autosomal recessive |
| **KREMEN1** | 99.75 % | 609898 | Ectodermal dysplasia 13, hair/tooth type, 617392 (3), Autosomal recessive |

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| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **KRT14** | 99.88 % | 148066 | Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001 (3), Autosomal recessive; Epidermolysis bullosa simplex 1C, localized, 131800 (3), Autosomal dominant; Dermatopathia pigmentosa reticularis, 125595 (3), Autosomal dominant; Epidermolysis bullosa simplex 1A, generalized severe, 131760 (3), Autosomal dominant; Naegeli-Franceschetti-Jadassohn syndrome, 161000 (3), Autosomal dominant; Epidermolysis bullosa simplex 1B, generalized intermediate, 131900 (3), Autosomal dominant |
| **KRT25** | 99.2 % | 616646 | Woolly hair, autosomal recessive 3, 616760 (3), Autosomal recessive |
| **KRT74** | 98.52 % | 608248 | Woolly hair, autosomal dominant, 194300 (3), Autosomal dominant; ?Hypotrichosis 3, 613981 (3), Autosomal dominant; ?Ectodermal dysplasia 7, hair/nail type, 614929 (3), Autosomal recessive |
| **KRT81** | 75.62 % | 602153 | Monilethrix 2, 621169 (3), Autosomal dominant |
| **KRT83** | 99.99 % | 602765 | Monilethrix 3, 621170 (3), Autosomal dominant; Erythrokeratodermia variabilis et progressiva 5, 617756 (3), Autosomal recessive |
| **LAMB3** | 99.71 % | 150310 | Epidermolysis bullosa, junctional 1B, severe, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional 1A, intermediate, 226650 (3), Autosomal recessive; Amelogenesis imperfecta, type IA, 104530 (3), Autosomal dominant |
| **LIPH** | 99.91 % | 607365 | Hypotrichosis 7, 604379 (3), Autosomal recessive; Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 (3), Autosomal recessive |
| **LPAR6** | 99.99 % | 609239 | Hypotrichosis 8, 278150 (3), Autosomal recessive; Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150 (3), Autosomal recessive |
| **LRP6** | 99.83 % | 603507 | {Coronary artery disease, autosomal dominant, 2}, 610947 (3), Autosomal dominant; Tooth agenesis, selective, 7, 616724 (3), Autosomal dominant; Exudative vitreoretinopathy 8, 621268 (3), Autosomal dominant |
| **LSS** | 99.85 % | 600909 | Hypotrichosis 14, 618275 (3), Autosomal recessive; Cataract 44, 616509 (3), Autosomal recessive; Alopecia-intellectual disability syndrome 4, 618840 (3), Autosomal recessive |
| **LTBP3** | 99.41 % | 602090 | Dental anomalies and short stature, 601216 (3), Autosomal recessive; Geleophysic dysplasia 3, 617809 (3), Autosomal dominant |
| **MBTPS2** | 99.85 % | 300294 | Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive |
| **MMP20** | 99.97 % | 604629 | Amelogenesis imperfecta, type IIA2, 612529 (3), Autosomal recessive |
| **MSX1** | 99.87 % | 142983 | Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 (3), Autosomal dominant; Ectodermal dysplasia 3, Witkop type, 189500 (3), Autosomal dominant; Orofacial cleft 5, 608874 (3), Autosomal dominant |
| **NECTIN1** | 99.95 % | 600644 | Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive |
| **NECTIN4** | 99.17 % | 609607 | Ectodermal dysplasia-syndactyly syndrome 1, 613573 (3), Autosomal recessive |
| **NFKB2** | 99.71 % | 164012 | Immunodeficiency, common variable, 10, 615577 (3), Autosomal dominant |
| **NFKBIA** | 99.86 % | 164008 | Ectodermal dysplasia and immunodeficiency 2, 612132 (3), Autosomal dominant |
| **ODAPH** | 99.87 % | 614829 | Amelogenesis imperfecta, type IIA4, 614832 (3), Autosomal recessive |
| **ORAI1** | 99.4 % | 610277 | Immunodeficiency 9, 612782 (3), Autosomal recessive; Myopathy, tubular aggregate, 2, 615883 (3), Autosomal dominant |
| **PADI3** | 98.72 % | 606755 | Uncombable hair syndrome, 191480 (3), Autosomal recessive |
| **PAX9** | 99.78 % | 167416 | Tooth agenesis, selective, 3, 604625 (3), Autosomal dominant |

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|---|---|---|---|
| **PEX1** | 98.36 % | 602136 | Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive |
| **PEX26** | 99.99 % | 608666 | Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive |
| **PEX6** | 99.89 % | 601498 | Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive |
| **PKP1** | 99.71 % | 601975 | Ectodermal dysplasia/skin fragility syndrome, 604536 (3), Autosomal recessive |
| **PRKD1** | 99.92 % | 605435 | Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant |
| **RELT** | 99.88 % | 611211 | Amelogenesis imperfecta, type IIIC, 618386 (3), Autosomal recessive |
| **RIPK4** | 99.68 % | 605706 | CHAND syndrome, 214350 (3), Autosomal recessive; Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650 (3), Autosomal recessive |
| **RMRP** | 98.59 % | 157660 | Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive |
| **ROGDI** | 98.83 % | 614574 | Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive |
| **RSPO4** | 96.41 % | 610573 | Anonychia congenita, 206800 (3), Autosomal recessive |
| **SLC10A7** | 99.96 % | 611459 | Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363 (3), Autosomal recessive |
| **SLC13A5** | 99.92 % | 608305 | Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 (3), Autosomal recessive |
| **SLC24A4** | 99.77 % | 609840 | [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 (3), Autosomal recessive; Amelogenesis imperfecta, type IIA5, 615887 (3), Autosomal recessive; [Skin/hair/eye pigmentation 6, blue/green eyes], 210750 (3), Autosomal recessive |
| **SMARCA2** | 99.82 % | 600014 | Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant; Blepharophimosis-impaired intellectual development syndrome, 619293 (3), Autosomal dominant |
| **SMARCA4** | 99.81 % | 603254 | Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant; ?Otosclerosis 12, 620792 (3), Autosomal dominant |
| **SMARCAD1** | 99.68 % | 612761 | Basan syndrome, 129200 (3), Autosomal dominant; Huriez syndrome, 181600 (3), Autosomal dominant; Adermatoglyphia, 136000 (3), Autosomal dominant |
| **SMARCB1** | 99.8 % | 601607 | Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant; Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant |
| **SMARCC2** | 99.06 % | 601734 | Coffin-Siris syndrome 8, 618362 (3), Autosomal dominant |
| **SMARCD1** | 99.36 % | 601735 | Coffin-Siris syndrome 11, 618779 (3), Autosomal dominant |
| **SMARCE1** | 99.26 % | 603111 | {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant |
| **SMOC2** | 98.78 % | 607223 | Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400 (3), Autosomal recessive |
| **SNRPE** | 98.33 % | 128260 | Hypotrichosis 11, 615059 (3), Autosomal dominant |
| **SOX11** | 99.95 % | 600898 | Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866 (3), Autosomal dominant |

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|---|---|---|---|
| **SOX4** | 99.14 % | 184430 | Intellectual developmental disorder with speech delay and dysmorphic facies, 618506 (3), Autosomal dominant |
| **SP6** | 99.97 % | 608613 | Amelogenesis imperfecta, type IK, 620104 (3), Autosomal dominant |
| **SREBF1** | 99.51 % | 184756 | Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016 (3), Autosomal dominant; Mucoepithelial dysplasia, hereditary, 158310 (3), Autosomal dominant |
| **SSUH2** | 99.77 % | 617479 | *No OMIM phenotypes* |
| **STIM1** | 99.89 % | 605921 | Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive |
| **TCHH** | 99.71 % | 190370 | ?Uncombable hair syndrome 3, 617252 (3), Autosomal recessive |
| **TGM3** | 99.96 % | 600238 | ?Uncombable hair syndrome 2, 617251 (3), Autosomal recessive |
| **TP63** | 99.8 % | 603273 | Premature ovarian failure 21, 620311 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; ADULT syndrome, 103285 (3), Autosomal dominant; Limb-mammary syndrome, 603543 (3), Autosomal dominant |
| **TSPEAR** | 99.99 % | 612920 | Tooth agenesis, selective, 10, 620173 (3), Autosomal recessive; ?Deafness, autosomal recessive 98, 614861 (3), Autosomal recessive; Ectodermal dysplasia 14, hypohidrotic/hair/tooth/nail type, 618180 (3), Autosomal recessive |
| **VPS4B** | 99.92 % | 609983 | *No OMIM phenotypes* |
| **WDR72** | 96.5 % | 613214 | Amelogenesis imperfecta, type IIA3, 613211 (3), Autosomal recessive |
| **WNT10A** | 99.77 % | 606268 | Schopf-Schulz-Passarge syndrome, 224750 (3), Autosomal recessive; Tooth agenesis, selective, 4, 150400 (3), Autosomal recessive, Autosomal dominant; Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980 (3), Autosomal recessive |
| **WNT10B** | 99.68 % | 601906 | Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant; Split-hand/foot malformation 6, 225300 (3), Autosomal recessive |

## 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.