Oslo universitetssykehus
Avdeling for medisinsk genetikk

Ektodermal dysplasi og hypodonti

Genpanel, versjon v02

* Enkelte genomiske regioner har lav eller ingen sekvensdekning ved eksomsekvensering. Dette skyldes at de har stor likhet med andre områder i genomet, slik at spesifikk gjenkjennelse av disse områdene og påvisning av varianter i disse områdene, blir vanskelig og upålitelig. Disse genetiske regionene har vi identifisert ved å benytte USCS segmental duplication hvor områder større enn 1 kb og ≥90% likhet med andre regioner i genomet , gjenkjennes (https://genome.ucsc.edu).

For noen gener ligger alle ekson i områder med segmentale duplikasjoner, så disse vil ikke bli analysert: **GJA1**

Vi gjør oppmerksom på at ved identifiseringav ekson oppstrøms for startkodon kan eksonnummereringen endres uten at transkript ID endres.

Avdelingens websider har en full oversikt over områder som er utelatt pga. segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
AXIN2	904	NM_004655.3		2-11	Oligodontia-colorectal cancer syndrome OMIM
CDH3	1762	NM_001793.4		1-16	Hypotrichosis, congenital, with juvenile macular dystrophy OMIM Ectodermal dysplasia, ectrodactyly, and macular dystrophy OMIM
CTSK	2536	NM_000396.3		2-8	Pycnodysostosis OMIM
EDA	3157	NM_001399.4		1-8	Tooth agenesis, selective, X-linked 1 OMIM Ectodermal dysplasia 1, hypohidrotic, X-linked OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
EDAR	2895	NM_022336.3		2-12	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive OMIM Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant OMIM
EDARADD	14341	NM_145861.2		1-6	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive OMIM Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant OMIM
EVC	3497	NM_153717.2		1-21	Ellis-van Creveld syndrome OMIM Weyers acrodental dysostosis OMIM
EVC2	19747	NM_147127.4		1-22	Ellis-van Creveld syndrome OMIM Weyers acrofacial dysostosis OMIM
FGF10	3666	NM_004465.1		1-3	LADD syndrome OMIM Aplasia of lacrimal and salivary glands OMIM
FGFR2	3689	NM_000141.4		2-18	Craniofacial-skeletal-dermatologic dysplasia OMIM LADD syndrome OMIM Beare-Stevenson cutis gyrata syndrome OMIM Apert syndrome OMIM
FGFR3	3690	NM_000142.4		2-18	LADD syndrome OMIM Crouzon syndrome with acanthosis nigricans OMIM
FLNA	3754	NM_001456.3		2-47	Melnick-Needles syndrome OMIM Terminal osseous dysplasia OMIM PubMed Frontometaphyseal dysplasia 1 OMIM Otopalatodigital syndrome, type I OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
GJA1	4274	NM_000165.4	2	2	Oculodentodigital dysplasia OMIM Erythrokeratodermia variabilis et progressiva OMIM Palmoplantar keratoderma with congenital alopecia OMIM Oculodentodigital dysplasia, autosomal recessive OMIM
GJB6	4288	NM_006783.4		3	Ectodermal dysplasia 2, Clouston type OMIM
GRHL2	2799	NM_024915.3		1-16	Ectodermal dysplasia/short stature syndrome OMIM
IFT122	13556	NM_052985.3	15-20	1-31	Cranioectodermal dysplasia 1 OMIM
LTBP3	6716	NM_001130144.2		1-28	Dental anomalies and short stature OMIM
MSX1	7391	NM_002448.3		1-2	Tooth agenesis, selective, 1, with or without orofacial cleft OMIM Ectodermal dysplasia 3, Witkop type OMIM
NFKBIA	7797	NM_020529.2		1-6	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency OMIM
OFD1	2567	NM_003611.2		1-23	Simpson-Golabi-Behmel syndrome, type 2 OMIM Orofaciodigital syndrome I OMIM
PAX9	8623	NM_006194.3		2-5	Tooth agenesis, selective, 3 OMIM
PITX2	9005	NM_153427.2		3-5	Peters anomaly OMIM Axenfeld-Rieger syndrome, type 1 OMIM
POLR1C	20194	NM_203290.2		1-9	Leukodystrophy, hypomyelinating, 11 OMIM
POLR3B	30348	NM_018082.5		1-28	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
PORCN	17652	NM_203475.2		2-15	Focal dermal hypoplasia OMIM
PVRL1	9706	NM_002855.4		1-6	Orofacial cleft 7 OMIM Cleft lip/palate-ectodermal dysplasia syndrome OMIM
PVRL4	19688	NM_030916.2		1-9	Ectodermal dysplasia-syndactyly syndrome 1 OMIM PubMed
RECQL4	9949	NM_004260.3		1-22	Rothmund-Thomson syndrome OMIM
RUNX2	10472	NM_001024630.3		2-9	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly OMIM Cleidocranial dysplasia, forme fruste, with brachydactyly OMIM Cleidocranial dysplasia, forme fruste, dental anomalies only OMIM Cleidocranial dysplasia OMIM
SMOC2	20323	NM_022138.2		1-13	Dentin dysplasia, type I, with microdontia and misshapen teeth OMIM
TBX3	11602	NM_005996.3		1-7	Ulnar-mammary syndrome OMIM
TP63	15979	NM_003722.4		1-14	Limb-mammary syndrome OMIM Hay-Wells syndrome OMIM Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3 OMIM Split-hand/foot malformation 4 OMIM ADULT syndrome OMIM Rapp-Hodgkin syndrome OMIM Orofacial cleft 8 OMIM
UBR1	16808	NM_174916.2		1-47	Johanson-Blizzard syndrome OMIM
WNT10A	13829	NM_025216.2		1-4	Schopf-Schulz-Passarge syndrome OMIM Odontoonychodermal dysplasia OMIM Tooth agenesis, selective, 4 OMIM