Oslo universitetssykehus
Avdeling for medisinsk genetikk

Iktyose

Genpanel, versjon v03

Tabellen er sortert på gennavn (HGNC gensymbol)
Navn på gen er iht. HGNC
Kolonnen **>x10** viser andel av genet som vi forventer blir lest med tilfredstillende kvalitet flere enn 10 ganger under sekvensering

* 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).

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
ABCA12	14637	NM_173076.2		1-53	Ichthyosis, congenital, autosomal recessive 4A OMIM Ichthyosis, autosomal recessive 4B (harlequin) OMIM
ABHD5	21396	NM_016006.4		1-7	Chanarin-Dorfman syndrome OMIM
ALOX12B	430	NM_001139.2		1-15	Ichthyosis, congenital, autosomal recessive 2 OMIM
ALOXE3	13743	NM_021628.2		2-16	Ichthyosis, congenital, autosomal recessive 3 OMIM
AP1S1	559	NM_001283.3		1-5	MEDNIK syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
CDSN	1802	NM_001264.4		1-2	Peeling skin syndrome 1 OMIM Hypotrichosis 2 OMIM
CERS3	23752	NM_178842.3		4-13	Ichthyosis, congenital, autosomal recessive 9 OMIM
CLDN1	2032	NM_021101.4		1-4	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis OMIM
CSTA	2481	NM_005213.3		1-3	Peeling skin syndrome 4 OMIM
CYP4F22	26820	NM_173483.3		3-14	Ichthyosis, congenital, autosomal recessive 5 OMIM
DSG1	3048	NM_001942.3		1-15	Keratosis palmoplantaris striata I, AD OMIM Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE OMIM
EBP	3133	NM_006579.2		2-5	Chondrodysplasia punctata, X-linked dominant OMIM MEND syndrome OMIM
ELOVL4	14415	NM_022726.3		1-6	Ichthyosis, spastic quadriplegia, and mental retardation OMIM
ERCC2	3434	NM_000400.3		1-23	Trichothiodystrophy 1, photosensitive OMIM
ERCC3	3435	NM_000122.1		1-15	Trichothiodystrophy 2, photosensitive OMIM
FLG	3748	NM_002016.1	3	2-3	Ichthyosis vulgaris OMIM
GJB2	4284	NM_004004.5		2	Keratitis-ichthyosis-deafness syndrome OMIM Hystrix-like ichthyosis with deafness OMIM Vohwinkel syndrome OMIM
GJB3	4285	NM_024009.2		2	Erythrokeratodermia variabilis et progressiva OMIM
GJB4	4286	NM_153212.2		2	Erythrokeratodermia variabilis with erythema gyratum repens OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
GJB6	4288	NM_006783.4		3	Ectodermal dysplasia 2, Clouston type OMIM
GTF2H5	21157	NM_207118.2		2-3	Trichothiodystrophy 3, photosensitive OMIM
KRT1	6412	NM_006121.3		1-9	Epidermolytic hyperkeratosis OMIM Palmoplantar keratoderma, nonepidermolytic OMIM Palmoplantar keratoderma, epidermolytic OMIM Keratosis palmoplantaris striata III OMIM Ichthyosis, cyclic, with epidermolytic hyperkeratosis OMIM Ichthyosis histrix, Curth-Macklin type OMIM
KRT10	6413	NM_000421.3		1-8	Epidermolytic hyperkeratosis OMIM Ichthyosis with confetti OMIM Ichthyosis, cyclic, with epidermolytic hyperkeratosis OMIM
KRT2	6439	NM_000423.2		1-9	Ichthyosis bullosa of Siemens OMIM
KRT9	6447	NM_000226.3		1-7	Palmoplantar keratoderma, epidermolytic OMIM
LIPN	23452	NM_001102469.1		1-9	Ichthyosis, congenital, autosomal recessive 8 OMIM
LOR	6663	NM_000427.2		2	Vohwinkel syndrome with ichthyosis OMIM
MBTPS2	15455	NM_015884.3		1-11	IFAP syndrome with or without BRESHECK syndrome OMIM ?Olmsted syndrome, X-linked OMIM Keratosis follicularis spinulosa decalvans, X-linked OMIM
NIPAL4	28018	NM_001099287.1		1-6	Ichthyosis, congenital, autosomal recessive 6 OMIM
PNPLA1	21246	NM_001145717.1		1-8	Ichthyosis, congenital, autosomal recessive 10 OMIM

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POMP	20330	NM_015932.5		1-6	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma OMIM
SLC27A4	10998	NM_005094.3		2-13	Ichthyosis prematurity syndrome OMIM
SLURP1	18746	NM_020427.2		1-3	Meleda disease OMIM
SNAP29	11133	NM_004782.3		1-5	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome OMIM
SPINK5	15464	NM_006846.3		1-33	Netherton syndrome OMIM
ST14	11344	NM_021978.3		1-19	Ichthyosis, congenital, autosomal recessive 11 OMIM
STS	11425	NM_000351.4		1-10	Ichthyosis, X-linked OMIM
TGM1	11777	NM_000359.2		2-15	Ichthyosis, congenital, autosomal recessive 1 OMIM
TGM5	11781	NM_201631.3		1-13	Peeling skin syndrome 2 OMIM
TRPV3	18084	NM_145068.3		2-18	Olmsted syndrome OMIM ?Palmoplantar keratoderma, nonepidermolytic, focal 2 OMIM