Oslo universitetssykehus
Avdeling for medisinsk genetikk

Epidermolysis bullosa (EB)

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: **KRT14**

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
ATP2A2	812	NM_001681.3		1-21	Darier disease OMIM
COL17A1	2194	NM_000494.3		2-56	Epidermolysis bullosa, junctional, non-Herlitz type OMIM Epidermolysis bullosa, junctional, localisata variant OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
COL7A1	2214	NM_000094.3		1-118	Epidermolysis bullosa dystrophica, AR OMIM Epidermolysis bullosa dystrophica, AD OMIM Transient bullous of the newborn OMIM EBD, localisata variant EBD, Bart type OMIM Epidermolysis bullosa, pretibial OMIM EBD inversa OMIM Epidermolysis bullosa pruriginosa OMIM
DSP	3052	NM_004415.2		1-24	Skin fragility-woolly hair syndrome OMIM Keratosis palmoplantaris striata II OMIM Epidermolysis bullosa, lethal acantholytic OMIM Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis OMIM
DST	1090	NM_001723.5		1-24	Epidermolysis bullosa simplex, autosomal recessive 2 OMIM
EXPH5	30578	NM_015065.2		1-6	Epidermolysis bullosa, nonspecific, autosomal recessive OMIM
FERMT1	15889	NM_017671.4		2-15	Kindler syndrome OMIM
ITGA3	6139	NM_002204.2		1-25	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital OMIM
ITGA6	6142	NM_000210.2		1-25	Epidermolysis bullosa, junctional, with pyloric stenosis OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
ITGB4	6158	NM_001005731.1		2-39	Epidermolysis bullosa, junctional, with pyloric atresia OMIM Epidermolysis bullosa, junctional, non-Herlitz type OMIM Epidermolysis bullosa of hands and feet 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	Ichthyosis, cyclic, with epidermolytic hyperkeratosis OMIM Ichthyosis with confetti OMIM Epidermolytic hyperkeratosis OMIM
KRT14	6416	NM_000526.4	1-8	1-8	Epidermolysis bullosa simplex, Weber-Cockayne type OMIM Epidermolysis bullosa simplex, recessive 1 OMIM Epidermolysis bullosa simplex, Koebner type OMIM Epidermolysis bullosa simplex, Dowling-Meara type OMIM Dermatopathia pigmentosa reticularis OMIM Naegeli-Franceschetti-Jadassohn syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
KRT5	6442	NM_000424.3		1-9	Epidermolysis bullosa simplex, Dowling-Meara type OMIM Epidermylysis bullosa simplex-MCR OMIM Dowling-Degos disease 1 OMIM Epidermolysis bullosa simplex-MP OMIM Epidermolysis bullosa simplex, Weber-Cockayne type OMIM Epidermolysis bullosa simplex, recessive 1 OMIM Epidermolysis bullosa simplex, Koebner type OMIM
LAMA3	6483	NM_000227.4		1-38	Epidermolysis bullosa, generalized atrophic benign OMIM Epidermolysis bullosa, junctional, Herlitz type OMIM
LAMB3	6490	NM_000228.2		2-23	Epidermolysis bullosa, junctional, non-Herlitz type OMIM Epidermolysis bullosa, junctional, Herlitz type OMIM
LAMC2	6493	NM_005562.2		1-23	Epidermolysis bullosa, junctional, non-Herlitz type OMIM Epidermolysis bullosa, junctional, Herlitz type OMIM
PKP1	9023	NM_001005337.2		1-13	Ectodermal dysplasia/skin fragility syndrome OMIM
PLEC	9069	NM_000445.4		2-33	Epidermolysis bullosa simplex with pyloric atresia OMIM Epidermolysis bullosa simplex with muscular dystrophy OMIM ?Epidermolysis bullosa simplex with nail dystrophy OMIM Muscular dystrophy, limb-girdle, type 2Q OMIM Epidermolysis bullosa simplex, Ogna type OMIM
TGM5	11781	NM_201631.3		1-13	Peeling skin syndrome 2 OMIM