

## 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  $\geq 90\%$  likhet med andre regioner i genomet, gjenkjennes (<https://genome.ucsc.edu>).

For noen gener ligger alle ekson i områder med segmentale duplikasjoner: **KRT14**

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

Avdelingens websider har en full oversikt over områder som er affisert av [segmentale duplikasjoner](#).

\*\* Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
<a href="#">ATP2A2</a>	<a href="#">812</a>	NM_001681.3		1-21	Darier disease <a href="#">OMIM</a>
<a href="#">COL17A1</a>	<a href="#">2194</a>	NM_000494.3		2-56	Epidermolysis bullosa, junctional, non-Herlitz type <a href="#">OMIM</a> Epidermolysis bullosa, junctional, localisata variant <a href="#">OMIM</a>

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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
<a href="#">ITGB4</a>	<a href="#">6158</a>	NM_001005731.1		2-39	Epidermolysis bullosa, junctional, with pyloric atresia <a href="#">OMIM</a> Epidermolysis bullosa, junctional, non-Herlitz type <a href="#">OMIM</a> Epidermolysis bullosa of hands and feet <a href="#">OMIM</a>
<a href="#">KRT1</a>	<a href="#">6412</a>	NM_006121.3		1-9	Epidermolytic hyperkeratosis <a href="#">OMIM</a> Palmoplantar keratoderma, nonepidermolytic <a href="#">OMIM</a> Palmoplantar keratoderma, epidermolytic <a href="#">OMIM</a> Keratosis palmoplantaris striata III <a href="#">OMIM</a> Ichthyosis, cyclic, with epidermolytic hyperkeratosis <a href="#">OMIM</a> Ichthyosis histrix, Curth-Macklin type <a href="#">OMIM</a>
<a href="#">KRT10</a>	<a href="#">6413</a>	NM_000421.3		1-8	Ichthyosis, cyclic, with epidermolytic hyperkeratosis <a href="#">OMIM</a> Ichthyosis with confetti <a href="#">OMIM</a> Epidermolytic hyperkeratosis <a href="#">OMIM</a>
<a href="#">KRT14</a>	<a href="#">6416</a>	NM_000526.4	<a href="#">1-8</a>	1-8	Epidermolysis bullosa simplex, Weber-Cockayne type <a href="#">OMIM</a> Epidermolysis bullosa simplex, recessive 1 <a href="#">OMIM</a> Epidermolysis bullosa simplex, Koebner type <a href="#">OMIM</a> Epidermolysis bullosa simplex, Dowling-Meara type <a href="#">OMIM</a> Dermatopathia pigmentosa reticularis <a href="#">OMIM</a> Naegeli-Franceschetti-Jadassohn syndrome <a href="#">OMIM</a>

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