

Full name:	Epidermolysis bullosa and bladder diseases (60 genes) - KUL
Version number:	v8_102022
Laboratory:	<u>Centrum Menselijke Erfelijkheid - KUL</u>
Created:	28 Jun 2019 - 14:08
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Related Diseases

- Acral peeling skin syndrome
- Aplasia cutis congenita
- Autosomal dominant generalized dystrophic epidermolysis bullosa
- Autosomal recessive generalized dystrophic epidermolysis bullosa, intermediate form
- Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form
- Centripetalis recessive dystrophic epidermolysis bullosa
- Dermatosparaxis Ehlers-Danlos syndrome
- Diffuse palmoplantar keratoderma with painful fissures
- Epidermolysis bullosa simplex due to BP230 deficiency
- Epidermolysis bullosa simplex due to exophilin 5 deficiency
- Epidermolysis bullosa simplex superficialis
- Epidermolysis bullosa simplex with pyloric atresia
- Epithelial recurrent erosion dystrophy
- Exfoliative ichthyosis
- Focal palmoplantar keratoderma with joint keratoses
- Fraser syndrome
- Hereditary hypotrichosis with recurrent skin vesicles
- Hypohidrotic ectodermal dysplasia with immunodeficiency
- Hypotrichosis simplex
- Hypotrichosis simplex of the scalp
- Intermediate generalized junctional epidermolysis bullosa
- Interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome
- Junctional epidermolysis bullosa with pyloric atresia

- Keratoderma hereditarium mutilans
- Kindler epidermolysis bullosa
- Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome
- Late-onset junctional epidermolysis bullosa
- Lethal acantholytic erosive disorder
- Localized dystrophic epidermolysis bullosa, acral form
- Localized dystrophic epidermolysis bullosa, nails only
- Localized dystrophic epidermolysis bullosa, pretibial form
- Localized junctional epidermolysis bullosa
- Monilethrix
- Musculocontractural Ehlers-Danlos syndrome
- Nephrotic syndrome-epidermolysis bullosa-sensorineural deafness syndrome
- Palmoplantar keratoderma-deafness syndrome
- Peeling skin syndrome type A
- Peeling skin syndrome type B
- Recessive dystrophic epidermolysis bullosa inversa
- Self-improving dystrophic epidermolysis bullosa
- Severe dermatitis-multiple allergies-metabolic wasting syndrome
- Striate palmoplantar keratoderma

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ADAMTS2</u>	95.00	0	NM_014244.5/ interpretable range CS1>95%
<u>ATP2A2</u>	95.00	0	NM_001681.4/ interpretable range CS1>95%
<u>ATP2C1</u>	95.00	0	NM_014382.5/ interpretable range CS1>95%
<u>CAST</u>	95.00	0	NM_001042440.5/ interpretable range CS1>95%
<u>CD151</u>	95.00	0	NM_004357.5/ interpretable range CS1>95%
<u>CDSN</u>	95.00	0	NM_001264.5/ interpretable range CS1>95%
<u>CHST14</u>	95.00	0	NM_130468.4/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CHST8</u>	95.00	0	NM_001127896.2/ interpretable range CS1>95%
<u>COL17A1</u>	95.00	0	NM_000494.4/ interpretable range CS1>95%
<u>COL5A1</u>	95.00	0	NM_000093.5/ interpretable range CS1>95%
<u>COL5A2</u>	95.00	0	NM_000393.5/ interpretable range CS1>95%
<u>COL7A1</u>	95.00	0	NM_000094.4/ interpretable range CS1>95%
<u>CSTA</u>	95.00	0	NM_005213.4/ interpretable range CS1>95%
<u>CSTB</u>	95.00	0	NM_000100.4/ interpretable range CS1>95%
<u>DSC3</u>	95.00	0	NM_024423.4/ interpretable range CS1>95%
<u>DSG1</u>	95.00	0	NM_001942.4/ interpretable range CS1>95%
<u>DSG2</u>	95.00	0	NM_001943.5/ interpretable range CS1>95%
<u>DSG3</u>	95.00	0	NM_001944.3/ interpretable range CS1>95%
<u>DSG4</u>	95.00	0	NM_177986.5/ interpretable range CS1>95%
<u>DSP</u>	95.00	0	NM_004415.4/ interpretable range CS1>95%
<u>DST</u>	95.00	0	NM_001723.7/ interpretable range CS1>95%
<u>EXPH5</u>	95.00	0	NM_015065.3/ interpretable range CS1>95%
<u>FERMT1</u>	95.00	0	NM_017671.5/ interpretable range CS1>95%
<u>FLG2</u>	95.00	0	NM_001014342.3/ interpretable range CS1>95%
<u>GJB2</u>	95.00	0	NM_004004.6/ interpretable range CS1>95%
<u>GRIP1</u>	95.00	0	NM_021150.4/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>IKBKG</u>	95.00	0	NM_001099857.5/ interpretable range CS1>95%
<u>ITGA3</u>	95.00	0	NM_002204.4/ interpretable range CS1>95%
<u>ITGA6</u>	95.00	0	NM_000210.4/ interpretable range CS1>95%
<u>ITGB4</u>	95.00	0	NM_001005731.3/ interpretable range CS1>95%
<u>JUP</u>	95.00	0	NM_002230.4/ interpretable range CS1>95%
<u>KLHL24</u>	95.00	0	NM_017644.3/ interpretable range CS1>95%
<u>KRT1</u>	95.00	0	NM_006121.4/ interpretable range CS1>95%
<u>KRT10</u>	95.00	0	NM_000421.5/ interpretable range CS1>95%
<u>KRT14</u>	95.00	0	NM_000526.5/ interpretable range CS1>95%
<u>KRT16</u>	95.00	0	NM_005557.4/ interpretable range CS1>95%
<u>KRT17</u>	95.00	0	NM_000422.3/ interpretable range CS1>95%
<u>KRT2</u>	95.00	0	NM_000423.3/ interpretable range CS1>95%
<u>KRT5</u>	95.00	0	NM_000424.4/ interpretable range CS1>95%
<u>KRT6A</u>	95.00	0	NM_005554.4/ interpretable range CS1>95%
<u>KRT6B</u>	95.00	0	NM_005555.4/ interpretable range CS1>95%
<u>KRT6C</u>	95.00	0	NM_173086.5/ interpretable range CS1>95%
<u>KRT9</u>	95.00	0	NM_000226.4/ interpretable range CS1>95%
<u>LAMA3</u>	95.00	0	NM_000227.6/ interpretable range CS1>95%
<u>LAMB3</u>	95.00	0	NM_000228.3/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>LAMC2</u>	95.00	0	NM_005562.3/ interpretable range CS1>95%
<u>MMP1</u>	95.00	0	NM_002421.4/ interpretable range CS1>95%
<u>NID1</u>	95.00	0	NM_002508.3/ interpretable range CS1>95%
<u>PKP1</u>	95.00	0	NM_001005337.3/ interpretable range CS1>95%
<u>PLEC</u>	95.00	0	NM_000445.5/ interpretable range CS1>95%
<u>PLOD3</u>	95.00	0	NM_001084.5/ interpretable range CS1>95%
<u>SERPINB8</u>	95.00	0	NM_198833.2/ interpretable range CS1>95%
<u>SLC39A4</u>	95.00	0	NM_130849.4/ interpretable range CS1>95%
<u>SPINK5</u>	95.00	0	NM_006846.4/ interpretable range CS1>95%
<u>TGM5</u>	95.00	0	NM_201631.4/ interpretable range CS1>95%
<u>TP63</u>	95.00	0	NM_003722.5/ interpretable range CS1>95%
<u>UROD</u>	95.00	0	NM_000374.5/ interpretable range CS1>95%
<u>UROS</u>	95.00	0	NM_000375.3/ interpretable range CS1>95%
<u>WNT10A</u>	95.00	0	NM_025216.3/ interpretable range CS1>95%