GENETIC TEST:
Epidermolysis bullosa (gene panel)

FULL NAME:	Epidermolysis bullosa (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Sequence analysis: entire coding region
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	19 Jul 2019 - 10:20
CHANGED:	07 Aug 2023 - 10:26
URL:	https://laboboeken.nexuzhealth.com/pboek/internet/GHB%20CME/14372

RELATED CONTENT

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
- Inherited epidermolysis bullosa
- 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 Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- ADAM metallopeptidase with thrombospondin type 1 motif 2
- ATPase sarcoplasmic/endoplasmic reticulum Ca2+ transporting 2
- ATPase secretory pathway Ca2+ transporting 1
- calpastatin
- CD151 molecule (Raph blood group)
- corneodesmosin

- carbohydrate sulfotransferase 14
- carbohydrate sulfotransferase 8
- collagen type XVII alpha 1 chain
- collagen type V alpha 1 chain
- collagen type V alpha 2 chain
- collagen type VII alpha 1 chain
- cystatin A
- cystatin B
- desmocollin 3
- desmoglein 1
- desmoglein 2
- desmoglein 3
- desmoglein 4
- desmoplakin
- dystonin
- exophilin 5
- FERM domain containing kindlin 1
- filaggrin 2
- gap junction protein beta 2
- glutamate receptor interacting protein 1
- inhibitor of nuclear factor kappa B kinase regulatory subunit gamma
- integrin subunit alpha 3
- integrin subunit alpha 6
- integrin subunit beta 4
- junction plakoglobin
- kelch like family member 24
- keratin 1
- keratin 10
- keratin 14
- keratin 16
- keratin 17
- keratin 2
- keratin 5

- keratin 6A
- keratin 6B
- keratin 6C
- keratin 9
- laminin subunit alpha 3
- laminin subunit beta 3
- laminin subunit gamma 2
- matrix metallopeptidase 1
- nidogen 1
- plakophilin 1
- plectin
- procollagen-lysine,2-oxoglutarate 5-dioxygenase 3
- serpin family B member 8
- solute carrier family 39 member 4
- serine peptidase inhibitor Kazal type 5
- transglutaminase 5
- tumor protein p63
- uroporphyrinogen decarboxylase
- uroporphyrinogen III synthase
- Wnt family member 10A

Related Gene Panels

- Epidermolysis bullosa and bladder diseases (60 genes) - KUL
