GENETIC TEST:
Ichthyosis (gene panel)

FULL NAME:	Ichthyosis (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):	4 - 6 months
CREATED:	19 Jul 2019 - 16:01
CHANGED:	19 Sep 2022 - 10:16
URL:	https://laboboeken.nexuzhealth.com/pboek/internet/GHB%20CME/14372

RELATED CONTENT

Related Diseases

- Acral peeling skin syndrome
- Acral self-healing collodion baby
- Acrodermatitis continua of Hallopeau
- Acrodermatitis enteropathica
- Acute neonatal citrullinemia type I
- Annular epidermolytic ichthyosis
- Arthrogryposis-renal dysfunction-cholestasis syndrome
- Autosomal dominant epidermolytic ichthyosis
- Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering
- Autosomal dominant generalized epidermolysis bullosa simplex, intermediate form
- Autosomal dominant generalized epidermolysis bullosa simplex, severe form
- Autosomal dominant palmoplantar keratoderma and congenital alopecia
- Autosomal recessive epidermolytic ichthyosis
- BRESEK syndrome
- Bathing suit ichthyosis
- Biotinidase deficiency
- CEDNIK syndrome
- CHILD syndrome
- CK syndrome
- Carbamoyl-phosphate synthetase 1 deficiency
- Classic maple syrup urine disease
- Combined immunodeficiency due to partial RAG1 deficiency
- Combined immunodeficiency with granulomatosis
- Congenital ichthyosiform erythroderma
- Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome
- Congenital reticular ichthyosiform erythroderma

- Cutaneous mastocytoma
- DITRA
- Diffuse palmoplantar keratoderma with painful fissures
- Dowling-Degos disease
- Epidermolysis bullosa simplex with circinate migratory erythema
- Epidermolysis bullosa simplex with mottled pigmentation
- Epidermolytic palmoplantar keratoderma
- Erythrokeratodermia variabilis
- Exfoliative ichthyosis
- Focal palmoplantar keratoderma with joint keratoses
- Generalized pustular psoriasis
- Harlequin ichthyosis
- Hidrotic ectodermal dysplasia
- Holocarboxylase synthetase deficiency
- Hypotrichosis simplex of the scalp
- Ichthyosis follicularis-alopecia-photophobia syndrome
- Ichthyosis hystrix of Curth-Macklin
- Ichthyosis-hypotrichosis syndrome
- Ichthyosis-prematurity syndrome
- Ichthyosis-short stature-brachydactyly-microspherophakia syndrome
- Intermediate maple syrup urine disease
- Intermittent maple syrup urine disease
- Isolated focal non-epidermolytic palmoplantar keratoderma
- KID syndrome
- KRT1-related diffuse nonepidermolytic keratoderma
- Keratoderma hereditarium mutilans
- Keratoderma hereditarium mutilans with ichthyosis
- Keratosis follicularis spinulosa decalvans
- Keratosis linearis-ichthyosis congenita-sclerosing keratoderma syndrome
- Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome
- Lamellar ichthyosis
- Late-onset citrullinemia type I
- Localized epidermolysis bullosa simplex

- MEDNIK syndrome
- MEND syndrome
- Menkes disease
- Monilethrix
- Multiple sulfatase deficiency
- Mutilating palmoplantar keratoderma with periorificial keratotic plaques
- Neonatal ichthyosis-sclerosing cholangitis syndrome
- Neonatal inflammatory skin and bowel disease
- Netherton syndrome
- Neutral lipid storage disease with ichthyosis
- Nodular urticaria pigmentosa
- Omenn syndrome
- Pachyonychia congenita
- Palmoplantar keratoderma-deafness syndrome
- Palmoplantar keratoderma-hereditary motor and sensory neuropathy syndrome
- Peeling skin syndrome type A
- Peeling skin syndrome type B
- Pityriasis rubra pilaris
- Plaque-form urticaria pigmentosa
- Progressive symmetric erythrokeratodermia
- Propionic acidemia
- Pustulosis palmaris et plantaris
- Pyruvate dehydrogenase E3 deficiency
- Recessive X-linked ichthyosis
- Refsum disease
- Rhizomelic chondrodysplasia punctata type 1
- Self-improving collodion baby
- Severe combined immunodeficiency due to DCLRE1C deficiency
- Severe combined immunodeficiency due to complete RAG1/2 deficiency
- Severe dermatitis-multiple allergies-metabolic wasting syndrome
- Sjögren-Larsson syndrome
- Striate palmoplantar keratoderma
- Superficial epidermolytic ichthyosis

- Syndromic recessive X-linked ichthyosis
- Thiamine-responsive maple syrup urine disease
- Trichothiodystrophy
- Typical urticaria pigmentosa
- Vitamin B12-unresponsive methylmalonic acidemia type mut-
- Vitamin B12-unresponsive methylmalonic acidemia type mut0
- X-linked dominant chondrodysplasia punctata
- Xeroderma pigmentosum-Cockayne syndrome complex

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- ATP binding cassette subfamily A member 12
- abhydrolase domain containing 5, lysophosphatidic acid acyltransferase
- ADAM metallopeptidase domain 17
- aldehyde dehydrogenase 3 family member A2
- arachidonate 12-lipoxygenase, 12R type
- arachidonate lipoxygenase 3
- adaptor related protein complex 1 subunit beta 1
- adaptor related protein complex 1 subunit sigma 1
- aspartic peptidase retroviral like 1
- argininosuccinate synthase 1
- ATPase copper transporting alpha
- branched chain keto acid dehydrogenase E1 subunit alpha
- branched chain keto acid dehydrogenase E1 subunit beta
- biotinidase
- Bruton tyrosine kinase

- calpain 12
- caspase recruitment domain family member 14
- caspase 14
- calpastatin
- corneodesmosin
- ceramide synthase 3
- carbohydrate sulfotransferase 8
- claudin 1
- carbamoyl-phosphate synthase 1
- cystatin A
- cathepsin B
- cytochrome P450 family 4 subfamily F member 22
- dihydrolipoamide branched chain transacylase E2
- DNA cross-link repair 1C
- dihydrolipoamide dehydrogenase
- desmoglein 1
- EBP cholestenol delta-isomerase
- ELOVL fatty acid elongase 1
- ELOVL fatty acid elongase 4
- ERCC excision repair 2, TFIIH core complex helicase subunit
- ERCC excision repair 3, TFIIH core complex helicase subunit
- filaggrin
- filaggrin 2
- glucosylceramidase beta 1
- gap junction protein alpha 1
- gap junction protein beta 2
- gap junction protein beta 3
- gap junction protein beta 4
- gap junction protein beta 6
- general transcription factor IIE subunit 2
- general transcription factor IIH subunit 5
- holocarboxylase synthetase
- interleukin 36 receptor antagonist

- 3-ketodihydrosphingosine reductase
- KIT proto-oncogene, receptor tyrosine kinase
- keratin 1
- keratin 10
- keratin 14
- keratin 16
- keratin 2
- keratin 5
- keratin 6C
- keratin 83
- keratin 9
- lipase family member N
- loricrin cornified envelope precursor protein
- membrane bound transcription factor peptidase, site 2
- methylmalonyl-CoA mutase
- M-phase specific PLK1 interacting protein
- NIPA like domain containing 4
- NAD(P) dependent steroid dehydrogenase-like
- propionyl-CoA carboxylase subunit alpha
- propionyl-CoA carboxylase subunit beta
- p53 apoptosis effector related to PMP22
- peroxisomal biogenesis factor 7
- phytanoyl-CoA 2-hydroxylase
- phosphatidylinositol glycan anchor biosynthesis class L
- patatin like phospholipase domain containing 1
- proteasome maturation protein
- recombination activating 1
- recombination activating 2
- ring finger protein 113A
- short chain dehydrogenase/reductase family 9C member 7
- serpin family B member 7
- serpin family B member 8
- sphingosine-1-phosphate lyase 1

- solute carrier family 25 member 13
- solute carrier family 27 member 4
- solute carrier family 30 member 2
- solute carrier family 39 member 4
- synaptosome associated protein 29
- serine peptidase inhibitor Kazal type 5
- sterol regulatory element binding transcription factor 1
- ST14 transmembrane serine protease matriptase
- steroid sulfatase
- sulfotransferase family 2B member 1
- sulfatase modifying factor 1
- T-box transcription factor 1
- transglutaminase 1
- transglutaminase 5
- transient receptor potential cation channel subfamily M member 4
- VPS33B interacting protein, apical-basolateral polarity regulator, spe-39 homolog
- VPS33B late endosome and lysosome associated

Related Gene Panels

- Ichthyosis and erythroderma (98 genes) - KUL
