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Full name:	Ichthyosis and erythroderma (98 genes) - KUL
Description:	https://firebasestorage.googleapis.com/v0/b/uz-laboboeken.appspot.com/o/GHB_CME%2Fgeno-dermatosen-NGS-specifieke%20genpanels%20(transcripten).pdf?alt=media&token=4ce21463-cf35-4d79-8bb2-c4b58310b259
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Laboratory:	Centrum Menselijke Erfelijkheid - KUL
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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 Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ABCA12	95.00	0	NM_173076.3
ABHD5	95.00	0	NM_016006.6
ADAM17	95.00	0	NM_003183.6
ALDH3A2	95.00	0	NM_000382.3
ALOX12B	95.00	0	NM_001139.3
ALOXE3	95.00	0	NM_021628.3
AP1B1	95.00	0	NM_001127.4
AP1S1	95.00	0	NM_001283.5
ASPRV1	95.00	0	NM_152792.4
ASS1	95.00	0	NM_000050.4
ATP7A	95.00	0	NM_000052.7
BCKDHA	95.00	0	NM_000709.4
BCKDHB	95.00	0	NM_183050.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
BTD	95.00	0	NM_001370658.1
BTK	95.00	0	NM_000061.3
CAPN12	95.00	0	NM_144691.4
CARD14	95.00	0	NM_024110.4
CASP14	95.00	0	NM_012114.3
CAST	95.00	0	NM_001042440.5
CDSN	95.00	0	NM_001264.5
CERS3	95.00	0	NM_178842.5
CHST8	95.00	0	NM_001127896.2
CLDN1	95.00	0	NM_021101.5
CPS1	95.00	0	NM_001875.5
CSTA	95.00	0	NM_005213.4
CTSB	95.00	0	NM_001908.5
CYP4F22	95.00	0	NM_173483.4
DBT	95.00	0	NM_001918.5
DCLRE1C	95.00	0	NM_001033855.3
DLD	95.00	0	NM_000108.5
DSG1	95.00	0	NM_001942.4
EBP	95.00	0	NM_006579.3
ELOVL1	95.00	0	NM_001256399.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ELOVL4	95.00	0	NM_022726.4
ERCC2	95.00	0	NM_000400.4
ERCC3	95.00	0	NM_000122.2
FLG	95.00	0	NM_002016.2
FLG2	95.00	0	NM_001014342.3
GBA1	95.00	0	NM_001005741.3
GJA1	95.00	0	NM_000165.5
GJB2	95.00	0	NM_004004.6
GJB3	95.00	0	NM_024009.3
GJB4	95.00	0	NM_153212.3
GJB6	95.00	0	NM_006783.5
GTF2E2	95.00	0	NM_002095.6
GTF2H5	95.00	0	NM_207118.3
HLCS	95.00	0	NM_000411.8
IL36RN	95.00	0	NM_012275.3
KDSR	95.00	0	NM_002035.4
KIT	95.00	0	NM_000222.3
KRT1	95.00	0	NM_006121.4
KRT10	95.00	0	NM_000421.5
KRT14	95.00	0	NM_000526.5

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
KRT16	95.00	0	NM_005557.4
KRT2	95.00	0	NM_000423.3
KRT5	95.00	0	NM_000424.4
KRT6C	95.00	0	NM_173086.5
KRT83	95.00	0	NM_002282.3
KRT9	95.00	0	NM_000226.4
LIPN	95.00	0	NM_001102469.2
LORICRIN	95.00	0	NM_000427.3
MBTPS2	95.00	0	NM_015884.4
MMUT	95.00	0	NM_000255.4
MPLKIP	95.00	0	NM_138701.4
NIPAL4	95.00	0	NM_001099287.2
NSDHL	95.00	0	NM_015922.3
PCCA	95.00	0	NM_000282.4
PCCB	95.00	0	NM_000532.5
PERP	95.00	0	NM_022121.5
PEX7	95.00	0	NM_000288.4
PHYH	95.00	0	NM_006214.4
PIGL	95.00	0	NM_004278.4
PNPLA1	95.00	0	NM_001145717.1

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
POMP	95.00	0	NM_015932.6
RAG1	95.00	0	NM_000448.3
RAG2	95.00	0	NM_000536.4
RNF113A	95.00	0	NM_006978.3
SDR9C7	95.00	0	NM_148897.3
SERPINB7	95.00	0	NM_003784.4
SERPINB8	95.00	0	NM_198833.2
SGPL1	95.00	0	NM_003901.4
SLC25A13	95.00	0	NM_014251.3
SLC27A4	95.00	0	NM_005094.4
SLC30A2	95.00	0	NM_001004434.3
SLC39A4	95.00	0	NM_130849.4
SNAP29	95.00	0	NM_004782.4
SPINK5	95.00	0	NM_006846.4
SREBF1	95.00	0	NM_004176.5
ST14	95.00	0	NM_021978.4
STS	95.00	0	NM_000351.7
SULT2B1	95.00	0	NM_177973.2
SUMF1	95.00	0	NM_182760.4
TBX1	95.00	0	NM_080647.1

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
TGM1	95.00	0	NM_000359.3
TGM5	95.00	0	NM_201631.4
TRPM4	95.00	0	NM_017636.4
VIPAS39	95.00	0	NM_022067.4
VPS33B	95.00	0	NM_018668.5