



# NIRK

NETWORK FOR ICHTHYOSSES AND RELATED KERATINIZATION DISORDERS

## Medical History & Clinical Findings Form

(One form for each affected individual)

Doctor's name: \_\_\_\_\_

Bar code: \_\_\_\_\_

Date of assessment: \_\_\_\_\_

Clinic/Hospital: \_\_\_\_\_

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Female: \_\_\_\_\_ Male: \_\_\_\_\_ Date of birth: \_\_\_\_\_

Affected relatives: yes \_\_\_\_\_ no \_\_\_\_\_ who: \_\_\_\_\_

Consanguineous family: yes \_\_\_\_\_ no \_\_\_\_\_ in which way: \_\_\_\_\_

Ethnic origin of the parents: \_\_\_\_\_

Please fill in or add family tree!

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### Diagnosis:

Confirmed \_\_\_\_\_

Suspected: \_\_\_\_\_

### Medical history:

Pregnancy: normal \_\_\_\_\_ complicated: \_\_\_\_\_

Preterm birth: no \_\_\_\_\_ yes: \_\_\_\_\_ Week of gestation: \_\_\_\_\_

Weight of birth: \_\_\_\_\_ g size: \_\_\_\_\_ cm

Course of labour: normal \_\_\_\_\_ delayed \_\_\_\_\_ Sectio caesarea \_\_\_\_\_

Photos at birth or during childhood: no \_\_\_\_\_ yes \_\_\_\_\_ when: \_\_\_\_\_

Collodion membrane: complete integument \_\_\_\_\_  
partial \_\_\_\_\_ location: \_\_\_\_\_  
none \_\_\_\_\_

Skin changes immediately after birth: yes \_\_\_\_\_ no \_\_\_\_\_ Since when: \_\_\_\_\_

Other clinical findings - specification of skin changes – at primary presentation (key words/aspects): \_\_\_\_\_

**Symptoms at birth or during the first weeks of life:**

Ectropion:	no	yes	
Visual/disorders of the eye:	no	yes	of which sort:
Eclabium:	no	yes	
Deformation of the ears:	no	yes	of which sort:
Defective hearing:	no	yes	of which sort:
Joint contractures:	no	yes	of which sort:
Skeletal anomalies:	no	yes	of which sort:
Hypotrichosis/hair anomalies:	no	yes	of which sort:
Mucosal anomalies:	no	yes	of which sort:
Nail anomalies:	no	yes	of which sort:
Skin infections:	no	yes	of which sort:
Blister formation:	no	yes	of which sort:
Neurological anomalies:	no	yes	of which sort:
Visceral anomalies:	no	yes	of which sort:
Other symptoms:			

**Clinical course to date:**

Better  
 Unchanged  
 Worsened  
 Varying  
 Relapsing/in phases

Ability to perspire/sweat:	fully normal	none	partial	Location: _____
Facial flush under physical stress:	no	yes		
Heat congestion:	no	yes	with collapse	no      yes
Keratinization disorder better in	winter	summer		no difference
Pruritus:	never	rarely	often	always
Other clinical findings:				

**Diagnostics so far:**

(date, clinic or institution, results in brief)

Histology, light microscopy of the skin:

Histology, ultrastructure/electron microscopy:

Microscopic hairanalysis:

Steroid sulfatase activity in plasma:

Transglutaminase-1 activity test:

Molecular diagnostics:

Other:

Photo documentation:

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**Therapy:**

(so far)	what with / where	app. period of treatment	effect ( + / 0 / - )
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Shower:

Tub bath:

Steam bath:

Mechanical keratolysis (e.g. pumice stone...):

Bland ointments/creams/oils:

Keratolytic ointments/creams (e.g. urea ...):

Other external methods (e.g. milk...):

Scalp therapy:

Systemic medicaments (e.g. retinoids):

Acute in-patient treatment:

In-patient rehabilitation:

Surgery:

Other:

**Clinical/dermatological examination / current findings:**

Date: \_\_\_\_\_

Body height: \_\_\_\_\_ cm

Weight: \_\_\_\_\_ kg

**Findings of scalp skin:**

no

yes (please tick details)

No scaling

Colour of scale:

white

light brown

dark brown

black

Quality of scale:

severe/crusted keratinplaques

moderate/complete

slight/not entire scalp

Erythema:

severe, dark red

moderate/pink red

slight redness/flush

no erythema

Folliculitis/Pyoderma

Other:

**Facial findings:**

no

yes (please tick details)

No scaling

Colour of scale:

white

light brown

dark brown

black

Quality of scale:

plaque-like

thick-lamellar

medium-lamellar

discrete, fine-lamellar

keratotic lichenification

Erythema:

severe, dark red

moderate, pink red

slight redness

no erythema

Follikulitis/pyoderma

Other:

Findings of chest and abdomen:

no

yes

(please tick details)

No scaling

Colour of scale:

white

light brown

dark brown

black

Quality of scale:

severe/crusted keratinplates

moderate/complete

slight

Erythema:

severe, dark red

moderate/pink red

slight redness

no erythema

Follikulitis/Pyoderma

Other:

Findings on back and buttocks:

no

yes

(please tick details)

No scaling

Colour of scale:

white

light brown

dark brown

black

Quality of scale:

plate-like

thick-lamellar

medium-lamellar

discrete, fine-lamellar

keratotic lichenification

Erythema:

severe, dark red

Moderate, pink red

slight redness/flush

no erythema

Folliculitis/pyoderma

Other:

Findings on arms:

no

yes

(please tick details)

Cubital fossa affected:

yes

no

No scaling

Colour of scale:

white

light brown

dark brown

black

Quality of scale:

severe/crusted keratin plaques

moderate/complete

slight

Erythema:

severe, dark red

moderate/pink red

slight redness

no erythema

Folliculitis/Pyoderma

Other:

Findings on legs:

no

yes

(please tick details)

Popliteal fossa affected:

yes

no

No scaling

Colour of scale:

white

light brown

dark brown

black

Quality of scale:

plaque-like

thick-lamellar

medium-lamellar

discrete, fine-lamellar

keratotic lichenification

Erythema:

severe, dark red

moderate, pink red

slight redness

no erythema

Folliculitis/pyoderma

Other:





**Biopsy in glutaraldehyde for ultrastructural analysis in Heidelberg, Dr. Hausser:**

no yes date:

Diagnosed by:

Result:

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**Biopsy for transglutaminase-1 activity testing (congenial ichthyosis) or analysis of LEKTI (Netherton syndrome) in Münster (fresh frozen, unfixed material for cryosectioning, avoid contact to formalin):**

no yes date:

Diagnosed by:

Result:

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**Biopsy for skin specific RNA-isolation in Cologne, Dr. Hennies (special transport medium):**

no yes date:

Diagnosed by:

Result:

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**EDTA-blood for molecular diagnostics in Cologne, Dr. Hennies (lamellar ichthyosis and others):**

no yes date:

Diagnosed by:

Result:

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**EDTA-blood for molecular diagnostics in Münster (Netherton syndrome and others):**

no yes date:

Diagnosed by:

Result:

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**EDTA-blood for steroid sulfatase activity testing in Göttingen:**

no

yes

date:

Diagnosed by:

Result:

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**EDTA-blood for molecular diagnostics in Cologne, Dr. Arin and Müller** (keratinization disorders with epidermolytic hyperkeratosis):

no

yes

date:

Diagnosed by:

Result:

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**EDTA-blood for molecular diagnostics in Marburg** (CHILD/Conradi-Hünermann-Happle syndrome):

no

yes

date:

Diagnosed by:

Result:

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**EDTA-blood for molecular diagnostics in Erlangen:**

no

yes

date:

Diagnosed by:

Result:

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**Microscopic hair shaft analysis:**

no

yes

date:

Diagnosed by:

Result:

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**Quantitative amino acid analysis of hair shafts, Aachen** (trichothiodystrophy):

no

yes

date:

Diagnosed by:

Result:

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**Heparin-blood for EBV-transformation, Cologne:**

no

yes

date:

Diagnosed by:

Result:

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**Other molecular investigations:**

no

yes

date:

If yes, of which kind: \_\_\_\_\_

Diagnosed by:

Result:

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**Molecular analysis of family members:**

no

yes

date:

Barcode	Gender	Date of birth	Relationship to the affected individual	Date of blood sample

Diagnosed by:

Result: