



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: _____

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!

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
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:

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 keratinplaques

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:

Other current findings:

Accentuated palmoplantar markings (hyperlinear hands/feet):		no	yes
Palmoplantar keratosis:	palms affected	no	yes
	If yes, in which way:		diffuse striated papulous
		other:	_____
	Soles affected	no	yes
	If yes, in which way:		diffuse Plaque-like papulous
		other:	_____
Hyperhidrosis:	palmar:	no	yes
	plantar:	no	yes
Blistering:	no	yes	location:
Alopecia:	no	yes	
		which type:	(diffuse, circumscribed, non/-scarred)
Hypotrichosis:	no	yes	
		which type:	(diffuse, circumscribed)
Hair shaft anomalies:	no	yes	which type:
Nail anomalies:	no	yes	
		clubbing	
		other:	_____
Visual / eye anomalies:	no	yes	which type:
Ektropion:	no	yes	
Ear deformities:	no	yes	which type:
Hearing disorder:	no	yes	which type:
Eclabium:	no	yes	

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

no yes date:

Diagnosed by:

Result:

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:

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

no yes date:

Diagnosed by:

Result:

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

no yes date:

Diagnosed by:

Result:

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

no yes date:

Diagnosed by:

Result:

EDTA-blood for steroid sulfatase activity testing in Göttingen:

no

yes

date:

Diagnosed by:

Result:

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

no

yes

date:

Diagnosed by:

Result:

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

no

yes

date:

Diagnosed by:

Result:

EDTA-blood for molecular diagnostics in Erlangen:

no

yes

date:

Diagnosed by:

Result:

Microscopic hair shaft analysis:

no

yes

date:

Diagnosed by:

Result:

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

no

yes

date:

Diagnosed by:

Result:

Heparin-blood for EBV-transformation, Cologne:

no

yes

date:

Diagnosed by:

Result:

Other molecular investigations:

no

yes

date:

If yes, of which kind: _____

Diagnosed by:

Result:

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: