Disease/OMIM	Gen	Chromosom	Exons	Labor
Acrodermatitis enteropathica OMIM 201100	SLC39A4	8q24.3	12	Maas
Acute intermittent Porphyria OMIM 176000	HMBS	11q23.3	15	Maas
Atrichia with Papular Lesions OMIM 209500, 203655	HR	8p21.2	19	Maas
Basal cell nevus syndrome OMIM 109400	PTCH	9q22.3	23	Erla Düs
Birt-Hogg-Dubé syndrome OMIM 135150	BHD	17p11.2	10	Maas
Bullous congenital ichthyosiform erythroderma Brocq (epidermolytic hyperkeratosis) OMIM 113800	KRT1, KRT10	12q13, 17q21-q22	8	Köln
Capillary malformation-arteriovenous malformation OMIM 608354	RASA1	5q13.3	25	Maas
Chondrodysplasia punctata 2 (Conradi-Hünermann-Happle syndrome) OMIM 302960	EBP	Xp11.23	4	Maas
Cowden syndrome OMIM 158350	PTEN	10q23.31	9	Düs
Cockayne syndrome complementation group A OMIM 216400	CS A	5q12.1	11	Tübi
Cockayne syndrome complementation group B OMIM 133540	CS B	10q11	18	Tübi
Comél-Netherton Syndrome OMIM 256500	SPINK5	5q32	33	Müns
Congenital Alopecia with Immuno-deficiency and Naildystrophy OMIM 601705	FOXN1 (=WHN)	17q11-q12	9	Maas
Congenital erythropoietic porphyria OMIM 263700	UROS	10q25.2-q26.3	10	Maas
Crouzon syndrome with acanthosis nigricans OMIM 134934	FGFR3	4p16.3	G380R*	Köln
Cutaneous Leiomyoma, familial, multiple OMIM 150800	FH	1q42.1	10	Maas
Cylindromatosis, familial OMIM 132700	CYLD	16q12-q13	20	Maas
Darier-White disease OMIM 124200	ATP2A2	12q23-q24.1	21	Maas
Doss-Porphyria OMIM 125270	ALAD	9q34	9	Maas
Dyskeratosis congenita (Zinsser-Cole-Engman) OMIM 305000, 127550	DKC1	Xq28	15	Maas

Dyskeratosis congenita (Scoggins type) OMIM 127550	TERC	3q21-28	1	Maas
Epidermolysis bullosa dystrophica OMIM 226600	COL7A1	3p21.3	118	Frei
Epidermolysis bullosa junctionalis, Herlitz / non Herlitz OMIM 226700; 226650	LAMB3	1q32	R635X* R42X* R635X* Q373X* 29insC*	Frei Erla
Epidermolysis bullosa junctionalis, non Herlitz OMIM 226650	COL17A1	10q24.3	56	Frei
Epidermolysis bullosa simplex OMIM 131760; 131800; 131900; 131960; 601001	KRT5; KRT14	12q13; 17q12-q21	9; 8	Frei Köln
Erythrokeratodermia variabilis/symmetrica OMIM 133200; 602036	GJB3; GJB4	1p35.1	1	Maas IBK Frei Kö-Gen
Erythropoietic protoporphyria OMIM 177000	FECH	18q21.3	11	Maas
Fabry disease OMIM 301500	GLA	Xq22	7	Maas
Familial keloids OMIM 148100	Unknown	Loci: 2q23 and 7p11	Unknown	Bos
Gardner Syndrome OMIM 175100	APC	5q21	15	Erlan
Hailey- Hailey disease OMIM 169600	ATP2C1	3q21-q24	27	Maas
Hemochromatosis OMIM 235200	HFE1	6p21.3	H63D*; C282Y*	Maas IBK Erlan
Hereditary Coproporphyria OMIM 121300	CPO	3q12	7	Maas
Hidrotic ectodermal dysplasia 2 (Clouston syndrome) OMIM 129500	GJB6	13q12	1	Maas IBK
Hypotrichosis simplex of scalp OMIM 146520	CDSN	6p21.3	2	Maas
Hypotrichosis-lymphedema-telangiectasia syndrome OMIM 607823	SOX18	20q13.33	2	Maas
Hypotrichosis-nail dystrophy	GJB2, GJB6 (GJB5)	13q12 (1p35.1)	1	Maas IBK
Ichthyosis, autosomal recessive, congenital OMIM 242100, 242300, 606545	TGM1, ALOX12B, ALOXE3	14q11.2; 17p13.2-p13.1		Kö-Gen
Ichthyosis bullosa of Siemens OMIM 146800	KRT2E	12q13.13	E493K*	Maas Köln
Incontinentia Pigmenti OMIM 308300	IKBKG	Xq28	9	Maas
Keratosis Palmoplantaris Striata I	DSG1	8q12.1	15	Maas

OMIM 148700				
KID/HID, PPK-deafness, Bart-Pumphrey, Vohwinkel, Nonsyndromic sensorineural deafness, and hypotrichosis-deafness syndromes OMIM 148210/602540, 148350, 149200, 124500, 220290/601544	GJB2	13q12	1	Maas IBK Frei Kö-Gen
Kindler Syndrome OMIM 173650	KIND1	20p13	15	Frei
Klippel-Trenaunay-Weber Syndrome OMIM 149000	VG5Q	5q13.3	E133K*	Maas Dres
Lamellar Ichthyosis 1 OMIM 242300	TGM1	14q11.2	14	Maas
Laminopathies: Dunnigan lipodystrophy, progeria Hutchinson-Gilford, Mandibulo-acral dysplasia, limb-girdle muscular dystrophy 1b, Emery-Dreifuss muscular dystrophy, dilated cardiomyopathy 1a, Charcot-Marie-Tooth 2b, Werner-like syndrome, restrictive dermopathy OMIM 151660, 176670, 148370, 159001, 181350, 115200, 605588, 277700, 275210 resp.	LMNA/C ZMPSTE24 PPARγ	1q21.2 1p34 3p25	12 10 8	Maas
Lymphedema-distichiasis, Meige yellow nail syndrome OMIM 153400	FOXC2	16q24.3	1	Maas
Mal de Meleda OMIM 148300	ARS Component B	8qter	3	Maas Frei Kö-Gen
Marfan Syndrome OMIM 154700	FBN1	15q21.1	65	Erlan
McGrath ectodermal dysplasia-skin fragility syndrome OMIM 604536	PKP1	1q32	13	Maas
Monilethrix OMIM 158000	KRTHB1, KRTHB3, KRTHB6	12q13	9	Maas Köln
Muckle-Wells syndrome, CINCA OMIM 191900, 607115	CIAS1	1q44	Exon 3*	Maas
Nonne-Milroy Lymphedema OMIM 153100	FLT4	5q35.3	31	Maas
Oculo-dento-digital dysplasia OMIM 164200	GJA1	6q22.31	1	Maas IBK
Oligodontia-colorectal cancer syndrome OMIM 608615	AXIN2	17q24	Exon 7*	Maas
Palmoplantar keratoderma, epidermolytic (Vorner type) OMIM 144200	KRT9	17q21.1	8	Kö-Gen
Papillon-Lefèvre/Haim-Munk syndrome	CTSC	11q14.2	8	Maas Kö-Gen

OMIM 245000, 245010				
Peeling Skin syndrome OMIM 270300	In preparation for publication			Maas
Porphyria cutanea tarda (hereditary; type II) OMIM 176100	UROD	1p34	10	Maas
Porphyria variegata OMIM 176200	PPOX	1q22-q23	13	Maas
Peutz-Jeghers-Syndrome OMIM 175200	STK 11	19p13.3	9	Maas
Pycnodysostosis OMIM 265800	CTSK	1q21.2	7	Maas
SADDAN Dysplasia OMIM 134934.0015	FGFR3	4p16.3	K650M*	Maas IBK
Sjogren-Larsson-Syndrome OMIM 270200	FALDH	17p11.2	10	Erlan
Teleangiectasia hereditaria I OMIM 187300	ENG	9q34.1	14	Maas
Teleangiectasia hereditaria II OMIM 600376	ACVRLK1	12q11-q14	9	Maas
Tricho-dento-osseous syndrome	DLX3	17q21.33	3	Maas
Trichothiodystrophy complementation group A OMIM 608780	TTD-A / GTF2H5	6q25.3	1	Tübi
Trichothiodystrophy complementation group B OMIM 133510	XP-B	2q21	14	Tübi
Trichothiodystrophy complementation group C OMIM 601675	XP-D	19q13.2-q13.3	17	Tübi
Vohwinkel syndrome (variant form); Camisa syndrome OMIM 604117	LOR	1q21	1	Maas IBK Kö-Gen
Werner syndrome	RECQL2	8p12-p11.2	35	Köln
Witkop syndrome OMIM 189500	MSX1	4p16.1	2	Maas
Xeroderma pigmentosa complementation group A OMIM 278700	XP-A	9q22.3	6	Tübi
Xeroderma pigmentosa complementation group B OMIM 133510	XP-B	2q21	14	Tübi
Xeroderma pigmentosa complementation group C OMIM 278720	XP-C	3p25	15	Tübi
Xeroderma pigmentosa complementation group D OMIM 278730	XP-D	19q13.2-q13.3	17	Tübi
Xeroderma pigmentosa complementation group E	XP-E	11p12-p11	9	Tübi

OMIM 278740				
Xeroderma pigmentosa complementation group F OMIM 278760	XP-F	16p13.3-p13.13	11	Tübi
Xeroderma pigmentosa complementation group G OMIM 278780	XP-G	13q33	15	Tübi
Experimental				
	DSG3	18q12.1	16	Maas
	IVL	1q21	1	Maas
	NARF	17q25.3	12	Maas
	FATP4	9q34.11	12	Maas
	CTSL	9q21		Maas
In preparation				
Basal cell nevus syndrome OMIM 109400	PTCH	9q22.3		Maas
CHILD syndrome OMIM 308050	NSDHL	Xq28		Maas
Chanarin-Dorfman disease OMIM 275630	CGI58	3p21	7	Maas
Disseminated superficial actinic porokeratosis type 1 OMIM 175900	SSH1	12q24.11		Maas
Epidermolysis bullosa with pyloric atresia OMIM 226730	ITGB4 ITGA6	17q25.1 2q31.1		Maas
Hypotrichosis, localized, recessive OMIM 607903	DSG4	18q12	16	Maas
Keratosis Palmoplantaris Striata II OMIM 125647 Carvajal-Huerta syndrome OMIM 605676	DSP	6p24	24	Maas
Kindler syndrome OMIM 173650	KIND1	20p12.3	14	Maas
Lamellar ichthyosis	Ichthyin	5q33	6	Maas
Naxos disease OMIM 601214	JUP	17q21	13	Maas
Nonbullous congenital ichthyosiform erythroderma 1 OMIM 242100	ALOXE3 ALOXB12	17p13.1 17p13.1	15 15	Maas
Palmoplantar keratoderma, epidermolytic (Vorner type) OMIM 144200	KRT1, KRT9	12q13, 17q21.1		Maas
Bullous Erythroderma Ichthyosiformis Congenita of Brocq	KRT1, KRT10	12q13, 17q21		Maas
Piebaldism, mastocytosis	KIT	4q12	21	Maas
Rothmund-Thomsen syndrome	RECQL4	8q24.3		Maas

OMIM 268400				
Weill-Marchesani syndrome OMIM 277600	ADAMTS10	19p13.3	24	Maas

Tabellarische Übersicht der Genodermatosen/hereditären Syndrome, die derzeit im Labor/in der Klinik eines Mitglieds der Arbeitsgruppe Genetik innerhalb der Arbeitsgemeinschaft Dermatologische Forschung molekulargenetisch untersucht werden können. Die Erkrankungen sind alphabetisch geordnet aufgelistet. Daneben findet sich die zugehörige OMIM-Nummer, das Gen-Symbol, die Anzahl der Exons des Gens/der Gene sowie das Labor, welches die DNA-Analyse durchführt. - Die einzelen Labore sollten vor dem evtl. Versandt von Proben kontaktiert werden, damit Art und Menge des Materials sowie die individuellen Kapazitäten mit dem Ziellabor abgesprochen werden können.

Abkürzungen/Symbole:

***** = nur einzelne Exone und/oder spezifische (Hotspot-)Mutationen werden untersucht

Symbole der Laboratorien/Kliniken (farbkodiert):

Maas Abteilung Dermatologie, Academisch Ziekenhuis Maastricht; Niederlanden

IBK Department of Medical Genetics and Molecular and Clinical Pharmacology, Division of Clinical Genetics, Medical University Innsbruck; Austria

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