

Molecular analyses for ichthyoses

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(Listed by countries)

Provided by the Ichthyosis Consensus Group

This list will be completed and updated from time to time (Internet-Link: <http://www.netzwerk-ichthyose.de/index.php?id=27&L=1>)

Country	Name and address of Laboratory	Contact	Genes	Comments	Accreditation
Austria	Department of Dermatology Innsbruck Medical University Anichstr. 35 A-6020 Innsbruck	Prof. Dr. Matthias Schmuth Matthias.schmuth@i-med.ac.at Dr. Robert Gruber R.gruber@i-med.ac.at	<i>FLG, GJB2, GJB3, GJB4, GJB6, LOR, STS,</i>	Research facilities, analysis upon request	No
France	Department of Genetics Necker hospital for sick children Tour Lavoisier 3 rd floor 149 rue de Sèvres 75015 Paris	Prof. Dr. Alain Hovnanian alain.hovnanian@inserm.fr Tél: +33 1 40 61 54 44	<i>SPINK5, FLG, TGM1 ABCA12, ALOXE3, ALOX12B, ICHTHYIN, CYP4F22, KRT1, KRT10, KRT6A, KRT16, KRT6B, KRT17, GJB2, GJB3, GJB4, GJB6, LOR, ATP2A2, ATP2C1</i>	Routine and research laboratory	Fully affiliated and accredited for molecular and prenatal diagnosis in patients and families
Germany	Labor Prof. Traupe Department of Dermatology University Hospital Münster Von-Esmarch-Str. 58 48149 Münster	Prof. Dr. Heiko Traupe traupeh@mednet.uni-muenster.de Dr. Vinzenz Oji ojiv@mednet.uni-muenster.de NIRK referral center: www.netzwerk-ichthyose.de http://www.netzwerk-ichthyose.de/fileadmin/nirk/uploads/technical_information_EN.pdf	<i>SPINK5, FLG, CDSN</i> Through cooperation with the partners from the <i>Network for ichthyoses and related keratinization disorders</i> (NIRK) we offer contact and organization (incl. DNA extraction) to analyze genes affected in ARCI, KPI, Vörner, EKV, CDPX2, CHILD, and others	Research facilities, analysis upon request	No

Germany	Dept. of Pediatrics Metabolic Lab Room 1D3 646 University of Göttingen Robert-Koch-Str. 40 37075 Göttingen	Prof. Dr. Dr. Robert Steinfeld ncl@med.uni-goettingen.de Phone: +49 (0) 551-3912833 See: http://www.paediatric2.med.uni-goettingen.de/38.htm	Enzyme activity test of steroid sulfatase/ arylsulfatase C (from peripheral leucocytes, EDTA blood analysis) <i>Sequencing of SUMF1</i>	Routine laboratory	Yes
Germany	Dept. of Dermatology, Venerology, and Allergology University of Goettingen Von-Siebold-Strasse 3 37075 Goettingen	Prof. Dr. Steffen Emmert semmert@gwdg.de Web-site: http://www.dermatologie.med.uni-goettingen.de/	<i>TTD-A, TTDN1, XPB, XPD</i> Functional tests for DNA repair deficiency <i>XPA, XPC, DDB1, DDB2, XPF, XPG, POL eta, CSA, CSB, SNAP29</i>	Research Lab	No
Germany	Dept. of Human Genetics Bahnhofstrasse 5 D-35033 Marburg Department of Dermatology Deutschhausstrasse 9 D-35033 Marburg	Prof. Dr. KH Grzeschik (Dept. of Humana Genetics) grzeschi@staff.uni-marburg.de Prof. A König (Dept. of Dermatology) koeniga@med.uni-marburg.de	<i>NSDHL, EBP, PORCN, MBTPS2</i>	Research facilities, analysis upon request; Outpatient clinics	No
Germany	University Hospital Freiburg Institute for Human Genetics Breisacher Str. 33 79106 Freiburg	Prof. Dr. Judith Fischer Web-site: http://www.uniklinik-freiburg.de/humangenetik/live/index.html	http://www.uniklinik-freiburg.de/humangenetik/live/index.html		Yes
Germany	EM Laboratory Department of Dermatology University Clinic Heidelberg Vossstr. 2 69115 Heidelberg	Dr. Ingrid Hausser ingrid.hausser@med.uni-heidelberg.de	Ultrastructural anaysis of skin biopsies as a tool for preclassification (marker for distinct candidate genes) or exclusion of candidate genes for disorders of cornification and other heritable skin disorders	Service diagnostic lab, international service (DIN ISO 9001:2000)	Yes
Israel	Laboratory of Molecular Dermatology Department of Dermatology Tel Aviv Sourasky Medical center 6, Weizmann street Tel Aviv 64239	Prof. Dr. Eli Sprecher Tel: 97236974287 Email: elisp@tasmc.health.gov.il	<i>ABCA12, ABHD5, ALOXE3, ALOX12B, CYP4F22, GJB2 GJB6, GJB3, GJB4, ichthyin, KRT1, KRT2, KRT10, KRT9, KRT14, KRT5, LIPN, LOR, SNAP29, SPINK5, ST14, TGM1, VPS33B</i>	Analysis upon request	No

Japan	The First Laboratory, Department of Dermatology Hokkaido University Graduate School of Medicine N15 W7, Kita-ku, Sapporo 060-8638	Hiroshi Shimizu, MD, PhD shimizu@med.hokudai.ac.jp Toshifumi Nomura, MD, PhD Nomura@huhp.hokudai.ac.jp	<i>ABCA12, FLG, TGM1, ABHD5, ALDH3A2, KRT1, KRT2, KRT10, GJB2, GJB6, GJB3, GJB4, EBP, LOXE3, ALOX12B, ichthyin, CYP4F22 LOR</i>	Research lab	No
Japan	Department of Dermatology and Kurume University Cutaneous Cell Biological Institute, Kurume University School of Medicine, 67 Asahimachi, Kurume, Fukuoka 830-0011	Takashi Hashimoto, MD hasimot@med.kurume-u.ac.jp Takahiro Hamada, MD Hamataka@med.kurume-u.ac.jp Shunpei Fukuda, MD Fukuda_shunpei@med.kurume-u.ac.jp	<i>FLG, STS, KRT1, KRT2, KRT10, TGM1, ABCA12, ALOXE3, ALOX12B, Ichthyin, CYP4F22, LIPN, LOR, GJB3, GJB4, CDSN, TGM5, SPINK5, MBTPS2, XPD</i>	Gene mutation analysis upon request	No
Japan	Department of Dermatology Nagoya University Graduate School of Medicine 65 Tsurumai-cho, Showa-ku, Nagoya 466-8550	Masashi Akiyama, MD, PhD makiyama@med.nagoya-u.ac.jp Kazumitsu Sugiura, MD, PhD kazusugi@med.nagoya-u.ac.jp Michihiro Kono, MD miro@med.nagoya-u.ac.jp	<i>ABCA12, FLG, TGM1, ABHD5, ALDH3A2, KRT1, KRT2, KRT10, GJB2, GJB6, GJB3, GJB4, EBP, ALOXE3, ALOX12B, ichthyin, CYP4F22, LOR, ATP2A2, ATP2C1</i>	Research lab	
The Netherlands	Experimental dermatology lab, dermatology, Maastricht University Medical Center, P Debijelaan 25, 6202 AZ Maastricht	Maurice van Steensel, MD PhD m.van.steensel@mumc.nl Michel van Geel, MD m.van.gee@mumc.nl	<i>FLG, ALOXE3, ALOX12B, ichthyin, TG1, TG5, loricrin, GJB2, GJB3, GJB4, GJB6, GJA1, EBP, NSDHL, ABHD5, FATP4, KRT1, KRT10, KRT2e</i>	Research lab	No, but analysis can be repeated in affiliated accredited lab if needed
USA	GeneDx CLIA approved molecular diagnostic laboratory 207 Perry Parkway Gaithersburg, Maryland 20877	genedx@genedx.com Gabriele Richard, MD, FACMG gabi@genedx.com	<i>ABCA12, ABHD5, ALDH3A2, ALOXE3, ALOX12B, FLG (select mutations) GJB2, GJB6, GJB3, GJB4, ichthyin, KRT1, KRT2, KRT9, KRT10, SPINK5, STS (deletion testing), TGM1.</i> Testing is also available for a large number of other disorders of cornification and other genetic skin disorders (see www.genedx.com for a complete test menu).	Full service diagnostic lab for clinical diagnosis, carrier testing, and prenatal diagnosis. International service.	Yes, fully accredited lab