

The diagram illustrates the network structure of the ERN ITHACA project. At the center is the **SGMG** (Swiss Genetic Metabolic Gene Group), labeled as the "coordinating core group". Surrounding SGMG are several key entities, each connected to a central hub (University Hospital Bern, HUG Genève, CHUV Lausanne, University Hospitals Zürich, or Universitätsspital Basel/University Childrens Hospital Basel) and further connected to various associated centers, physicians, and organizations.

Legend:

- Ref center (Blue line)
- RC-ass. C/P (Light blue line)
- Ass.-center (Green line)
- Ass. experts (Light green line)
- Pat. organisations (Orange line)
- Prof. organisations (Red line)

Key Entities and Connections:

- SGMG (coordinating core group)** is connected to:
 - University Hospital Bern** (Ref center)
 - HUG Genève** (Ref center)
 - CHUV Lausanne** (Ref center)
 - University Hospitals Zürich** (Ref center)
 - Universitätsspital Basel/University Childrens Hospital Basel** (Ref center)
- University Hospital Bern** is connected to:
 - MaRaVal** (Pat. organisations)
 - ProRaris** (Pat. organisations)
 - Kantonsspital Luzern** (Ref center)
 - HVS / ZIS Sion** (Ass.-center)
 - AEMO** (Pat. organisations)
 - Spina bifida Hydrocephalus** (Pat. organisations)
 - KS Fribourg** (Ass.-center)
 - Angelman** (Pat. organisations)
 - CEMARAD** (Ass.-center)
 - Williams-Beuren** (Pat. organisations)
 - Phelan McDermid** (Pat. organisations)
 - Klinik Lengg Zürich** (Ass.-center)
 - Noonan-Syndrom** (Pat. organisations)
 - Epi-Suisse** (Pat. organisations)
 - Kantonsspital Winterthur** (Ref center)
 - University Hospitals Zürich** (Ref center)
- HUG Genève** is connected to:
 - associated centers** (RC-ass. C/P)
 - associated physicians** (RC-ass. C/P)
 - CHUV Lausanne** (Ref center)
- CHUV Lausanne** is connected to:
 - associated centers** (RC-ass. C/P)
 - associated physicians** (RC-ass. C/P)
 - University Hospitals Zürich** (Ref center)
- University Hospitals Zürich** is connected to:
 - associated centers** (RC-ass. C/P)
 - associated physicians** (RC-ass. C/P)
 - Universitätsspital Basel/University Childrens Hospital Basel** (Ref center)
- Universitätsspital Basel/University Childrens Hospital Basel** is connected to:
 - associated centers** (RC-ass. C/P)
 - associated physicians** (RC-ass. C/P)
 - University Hospital Bern** (Ref center)
- University Hospital Bern** is also connected to:
 - SSHID** (Prof. organisations)
 - Kindserspital St. Gallen** (Ass.-center)
 - Kantonsspital Aarau** (Ref center)
 - Epilipsie-Liga** (Prof. organisations)
 - Rett** (Pat. organisations)
 - MGR** (Pat. organisations)
 - EOC Ticino** (Ass.-center)

ref. center
 RC-ass. center
 ass. center
 ass. experts

Modified map from: <https://www.kosekschweiz.ch/anlaufstellen/zentrenseltenkrankheiten>

SwissITHACA member list

Candidate Reference Centers

Zentrum für seltene Entwicklungsstörungen Bern (ITHACA) / University Hospital Bern	http://www.humangenetik.insel.ch/de/ Prof. Dr. Dr. med. Christiane Zweier, Humangenetik humangenetik@insel.ch
Zentrum für seltene Entwicklungsstörungen – Ithaca Basel / Universitätsspital Basel/University Childrens Hospital Basel	Prof. Dr. med. Isabel Filges, Medizinische Genetik, USB Prof. Dr. med. Alexandre Datta, Neuropädiatrie, UKBB rarediseasesbasel@ukbb.ch
Referenzzentrum für seltene Entwicklungsstörungen Zürich (ITHACA Zürich) / University of Zürich and its associated Hospitals (plus associated centers KS Graubünden and KS Winterthur)	<u>ITHACA Zürich</u> Contact: Tanja Frey, med. pract.; tanja.frey@medgen.uzh.ch <u>KS Graubünden:</u> www.ksgr.ch ; marianne.meuli@ksgr.ch <u>KS Winterthur:</u> Sozialpädiatrisches Zentrum, Entwicklungspädiatrie und Neuropädiatrie; ep.spz@ksw.ch
Centre de référence suisse des maladies rares du développement / HUG Genève	https://www.hug.ch/medecine-genetique Chef de service : Marc.Abramowicz@hug.ch Médecins adjoints : Loredana.dAmato-Sizonenko@hug.ch Philippe.KhauvanKien@hug.ch Chef.fe.s de clinique : Rosalinda.Giannini@hug.ch Henri.Margot@hug.ch Roxane.vanHeurck@hug.ch Geraldine.vanWinckel@hug.ch Médecins internes : Sebastian.Suchet@hug.ch Omar.Zgheib@hug.ch Conseillère en génétique : Enora.LeTallec@hug.ch
Centre des malformations congénitales et/ou déficiences intellectuelles rares - ITHACA CHUV / CHUV Lausanne	Service de médecine génétique - Accueil - CHUV Contact email: gen@chuv.ch Responsable: Dr Jean-Marc Good
Referenzzentrum für seltene Entwicklungsstörungen Luzern (ITHACA Luzern)	Dr. Céline Gerber, celine.gerber@luks.ch ; Genetische Beratung, Luzerner Kantonsspital, Spitalstrasse, 6000 Luzern, sekretariatgenetik@luks.ch und Genetische

	Beratungsstelle Genetica, Falkenstrasse 14, 8024 Zürich, genetischeberatung@hin.ch
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Network Associated Centers

Spital Wallis (HVS) – Zentralinstitut der Spitler (ZIS), Sion	https://www.hopitalduvalais.ch/disciplines-medicales/disciplines-de-a-a-z/genetique Dr Claudine Rieubland, spcialiste FMH en gntique en mdicale ich.genetique@hopitalvs.ch
Ostschweizer Kinderspital St. Gallen	
Ente Ospedaliore Cantonale (EOC), Ticino	https://www.eoc.ch/professionisti-e-ricerca/medici-inviati/genetica-medica.html Dr.med. Silvia B. Maitz Servizio di Genetica Medica Geneticamedica.oil@eoc.ch
CEMARAD (Centre Expert des Maladies Rares du Dveloppement), Geneva	
Fribourg Hospital	

Network Associated Experts

Schweizerisches Epilepsie Zentrum / Klinik Lengg AG	
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Network Associated Professional Organisations

Schweizerische Gesellschaft fr Gesundheit bei Menschen mit intellektuellen Entwicklungsstrungen (SSHID = Swiss Society for Health in Intellectual Disability)	www.sshid.info
Schweizerische Epilepsie-Liga	www.epi.ch

Patient Organisations/Support Groups

Verein Noonan-Syndrom Schweiz	www.noonansyndrom.ch info@noonansyndrom.ch
Angelman Verein Schweiz	www.angelman.ch Melanie Della Rossa verein@angelman.ch
Frderverein fr Kinder mit Seltenen Krankheiten KMSK	www.kmsk.ch

AEMO (Association Enfance et Maladies Orphelines)	https://aemo.ch/
MaRaVal (maladies rares valais)	www.maraval.ch ; contact@maraval.ch Contact: Christine de Kalbermatten cdekalbermatten@maraval.ch Phone 027 321 27 47
MGR (Associazione Malattie Genetiche Rare Svizzera italiana)	info@malattierare.ch
RETT Cure Research Institute	
Phelan McDermid Gesellschaft e.V.	https://www.22q13.info/das-pmd-syndrom/
Leben mit Spina bifida und Hydrocephalus Schweiz	https://spina-hydro.ch/ Hanny Müller-Kessler SBH Schweiz (geschaeftsstelle@spina-hydro.ch)
Epi-Suisse, Schweizerischer Verein für Epilepsie	www.epi-suisse.ch Kontakt: Dominique Meier, Geschäftsstelle info@epi-suisse.ch
Williams Beuren Syndrom Schweiz	www.williams-syndrome.ch
KAT6B Switzerland	https://kat6b-suisse.org