



UEMS - EUROPEAN UNION OF MEDICAL SPECIALISTS

MULTIDISCIPLINARY JOINT COMMITTEE ON RARE AND UNDIAGNOSED DISEASES

UNDIAGNOSED DISEASE NETWORK INTERNATIONAL (UDNI) STRATEGIC PARTNER OF MJC RUD

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www.iss.it/web/iss-en/rare-diseases



UDNI Background



- Established in 2014
- The major aims is to ***provide diagnosis to patients*** and to ***foster research on novel diseases***, their mechanisms, and their pathways.
- UDNI involved originally **Austria, Bulgaria, Hungary, Italy, United States and Sweden.**
- A continuously growing number of countries is joining the network



www.iss.it/web/iss-en/rare-diseases





Undiagnosed
Diseases Network
INTERNATIONAL

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[Home](#) [Network](#) [Participants](#) [Platform resources](#) [Dissemination](#) [News and Events](#) [Patient area](#)



First Conference of Undiagnosed Diseases Network International
29-30 September 2014, Rome



How to apply

Requirements for Online Application
[VIEW PAGE](#)



Newsletter

To know project achievements and events
[SIGN UP](#)



UDNI Countries

[NCBI](#) [Resources](#) [How To](#)

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National Institutes of Health

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Format: Abstract

[Mol Genet Metab](#). 2015 Dec;116(4):223-5. doi: 10.1016/j.ymgme.2015.11.003. Epub 2015 Nov 5.

Undiagnosed Diseases Network International (UDNI): White paper for global actions to meet patient needs.

Taruscio D¹, Groft SC², Cederroth H³, Melegh B⁴, Lasko P⁵, Kosaki K⁶, Bavnam G⁷, McCray A⁸, Gahl WA⁹.

[Author information](#)



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NATIONAL CENTRE
RARE DISEASES

**UDNI:
the first Conference in
Rome (2014)
and
the we published the
white paper (2015)**

UDNI AIMS

- ✓ **To improve the level of diagnosis and care** for patients with UND through the development of common protocols designed by a large community of investigators.
- ✓ To facilitate research into the etiology of undiagnosed diseases, **by collecting and sharing standardized, high-quality clinical and laboratory data, including genotyping, phenotyping, and documentation of environmental exposures.**
- ✓ To create **an integrated and collaborative community across multiple Countries** and among laboratory and clinical investigators prepared to investigate the pathophysiology of these newly recognized and rare diseases.
- ✓ **TO SHARE: Best practices, protocols, consents, phenotypes, genomic data, functional analyses and models, knowledge management tools**



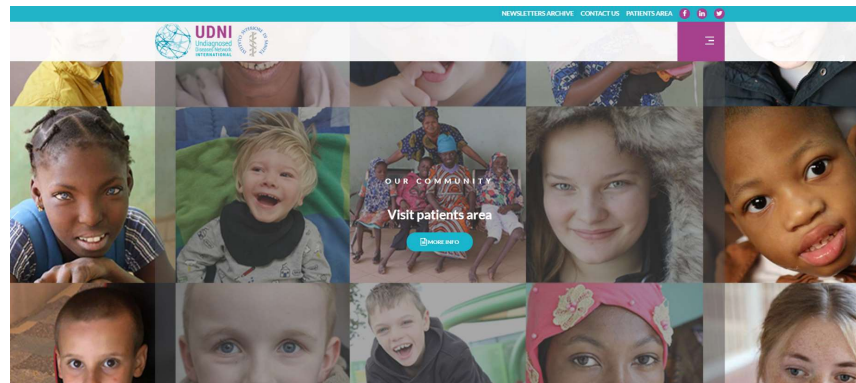
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Governance

- UDNI is partially modelled after the **NIH UDN** and its Manual of Operations, a handbook that details the network's research procedures and facilitates consistency across all participating institutions.
- The UDNI consists of a **Governing Board** and **Members**.
- **Specific Committees** and **Working Groups** are established to work on specific issues of interest

- A dedicated web site

www.udninternational.org



www.iss.it/web/iss-en/rare-diseases

UDNI PRINCIPLES		UDNI IMPLEMENTATION APPROACHES
1	Engage centers with experience in undiagnosed and rare diseases	Open membership to clinical and non-clinical members from around the world who are conducting undiagnosed diseases research.
2	Foster a collaborative research community	Hold periodic face-to-face meetings in international venues.
3	Establish a cooperative governance structure	Develop a process for membership involvement in key UDNI-wide decisions.
4	Design common research procedures	Clinical and basic research adheres to specific procedures (Applications, Evaluations, Sequencing).
5	Provide a high-quality patient evaluation	Establish UDNI-wide clinical and phenotypic evaluation methods and guidelines for patient acceptance.
6	Provide a high-quality patient experience	Acquire feedback from patients
7	Collect data in accordance with recognized data standards	Annotate clinical data with Human Phenotype Ontology. Follow international standards for human genomic data sharing.
8	Protect patient data	Develop and use accepted informed consent procedures, according to international standards for protecting research participants.
9	Expect broad data sharing	Share data both within and outside of the UDNI partner (Data Sharing).
10	Stimulate dissemination of research results	Publish research results. Develop policies for UDNI-wide publications.
11	Ensure a high-functioning network	Establish formal procedures for monitoring the progress of the UDNI.



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1st UDNI conference September 2014, Rome



2nd UDNI conference June 2015, Budapest



3rd UDNI conference February 2016, Wien



4th UDNI conference November 2016, Tokyo



5th UDNI conference August 2017, Stockholm



6th UDNI conference June 2018, Naples



7th UDNI conference April 2019, Dehli, India



8th UDNI conference February 2020, The Netherland



9th UDNI conference April 2021, Mayo Clinic, Minnesota USA



Turin 10th UDNI conference, 31 January – 1 February, 2022



UDNI WORKING GROUP



- Program Committee Group
- Communication Group
- Membership Committee
- Functional Study Group
- Data Sharing Group

- Patient Engagement Group
- Genetic Counseling Group
- **Educational Group**
- Developing Countries Group
- *Diagnostic Group (in progress)*

https://www.udninternational.org/schede-10-groups_with_udn_programs_activities_ongoing

UDNI specific Groups

UDNI Program Committee Group: linking the Program to a scientific meeting is considered beneficial

UDNI Communication Group: to coordinate UDNI information transfer; to provide information on how to apply to the UDNI, to coordinate and publish the UDNI newsletter.

UDNI Membership Committee: reviews all applications for membership and accept new members on a rolling basis.

UDNI Functional Study Group: to help clinicians find research collaborators and to submit cases to experts for opinions

UDNI Data Sharing Group: to make recommendations regarding UDNI sharing that involves rules of submission, storage and access.

UDNI Patient Engagement Group: to provide a patient perspective to issues like data sharing and to offer clarity and transparency.

UDNI Genetic Counseling Group: to address the genetic counseling needs of patients and their families who participate in an Undiagnosed Diseases Program.

UDNI Educational Group: to address educational issues related to undiagnosed diseases

UDNI Developing Countries Group: to address the issues (difficulties and opportunities) for providing diagnoses to patients with undiagnosed and rare diseases in Developing Countries



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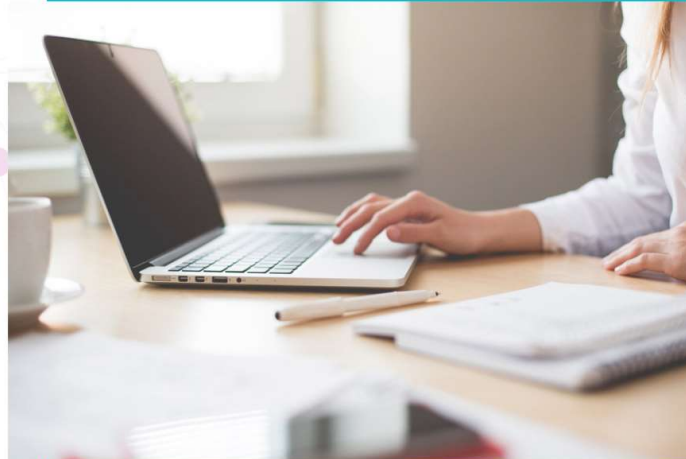
HOW TO APPLY

HOW TO BECOME AN UDNI MEMBER (FOR CLINICIANS AND RESEARCHERS)

Membership is opened to clinical investigators who serve undiagnosed disease patients from all countries. Before applying to UDNI as a member, please verify the following Requirements for Online Application.

- UDNI specific goals +
- Requirement for online application: +
- Sharing cases with UDNI at international level: +

APPLY AS A MEMBER



On line application for clinicians, researchers and patients

www.udninternational.org



HOW TO APPLY

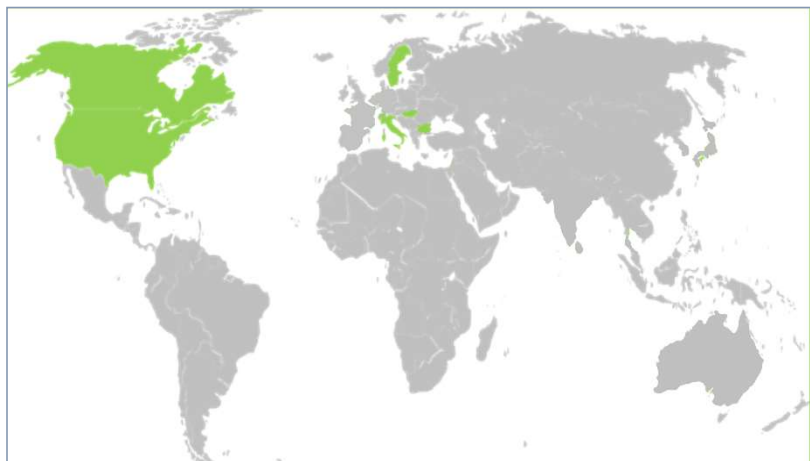
HOW TO BECOME AN UDNI PATIENT OR PATIENT'S ASSOCIATION MEMBER

If you decide to apply to UDNI as a patient, you will need to answer a series of brief questions through our application portal.

Referral letter from a primary licensed healthcare provider +

APPLY AS A PATIENT

APPLY AS AN ASSOCIATION



UDNI Participating Countries (2014 – 2021)

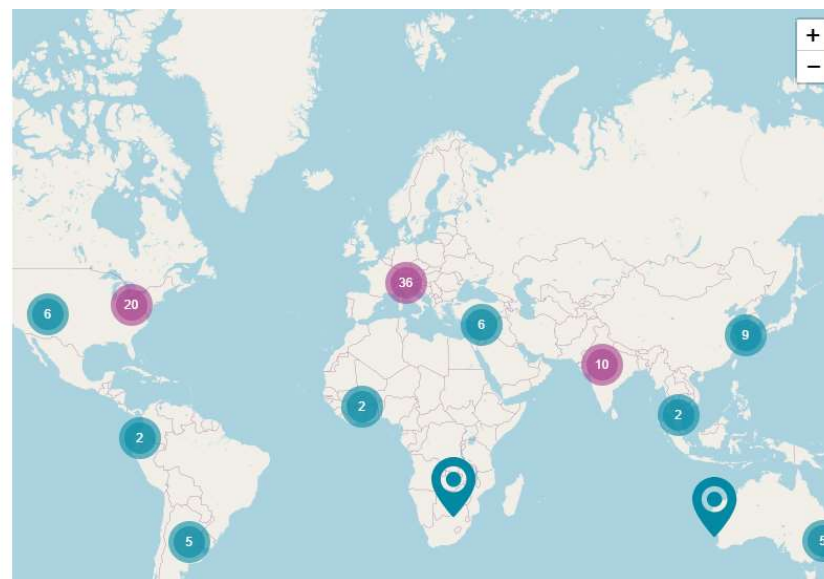
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UDNI MEMBER COUNTRIES

COUNTRIES PARTICIPATING TO THE UDNI

For each UDNI participating Country, please, point your mouse cursor in order to check coordinates of the involved Structure and Local Responsible for the program

[LIST OF UDNI COUNTRIES](#)




www.iss.it/web/iss-en/rare-diseases




ARGENTINA JAPAN
AUSTRALIA KOREA
AUSTRIA MALI
BELGIUM MEXICO
BELGUIM NEW ZELAND
BRAZIL PAKISTAN
BULGARIA PHILIPPINES
CANADA SAUDI ARABIA
CHILE SERBIA
CHINA SINGAPORE
ECUADOR SOUTH AFRICA
FRANCE SPAIN
GEORGIA SRI LANKA
GERMANY SWEDEN
GHANA SWITZERLAND
HONG KONG THAILAND
HUNGARY THE
INDIA NETHERLANDS
ISRAEL TURKEY
ITALY USA

Countries: 10 in 2014 > 39 in 2021






Molecular Genetics and Metabolism

Available online 17 January 2020
In Press, Journal Pre-proof



The Undiagnosed Diseases Network International: Five years and more!

D. Taruscio ^a  , G. Baynam ^{b, c}, H. Cederroth ^d, S.C. Groft ^e, E.W. Klee ^f, K. Kosaki ^g, P. Lasko ^{h, i}, B. Melegh ^j, O. Riess ^k, M. Salvatore ^a, W.A. Gahl ^{l, m}

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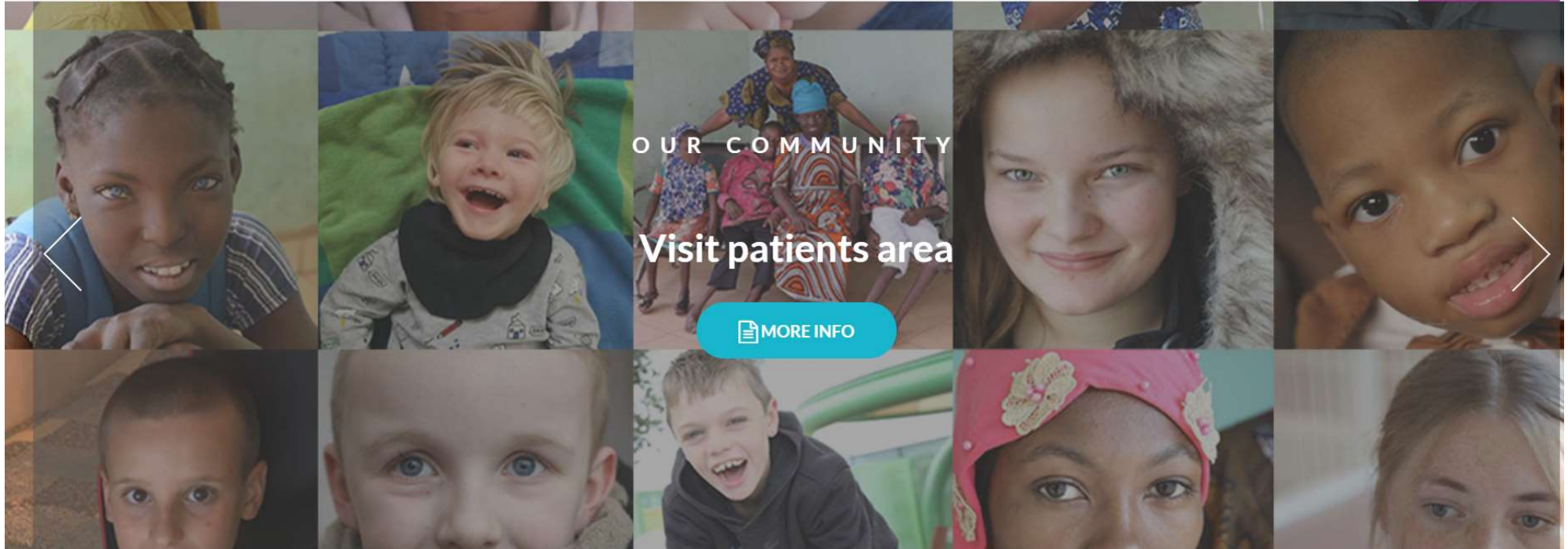
<https://doi.org/10.1016/j.ymgme.2020.01.004> [Get rights and content](#)

UDNI PATIENTS ASSOCIATIONS

NEWSLETTERS ARCHIVE CONTACT US PATIENTS AREA



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NATIONAL CENTRE
RARE DISEASES

UDNI PATIENTS ASSOCIATIONS

www.udninternational.org

- The Wilhelm Foundation
- NORD - National Organization for Rare Disorders
- Comitato "I Malati Invisibili" Onlus
- CORD - Chinese Organization for Rare Disorders
- FEDER - Federation Espanola de Enfermedades Raras
- EURORDIS - Rare Diseases Europe
- VSOP
- D'Genes - Association of rare diseases of Murcia
- Rare & Undiagnosed Network (RUN)
- Pancyprian Association for Rare Genetic Diseases "Unique SMILES"
- Organization of Rare Disease India
- Indo US Organization for Rare Diseases
- Syndrome Without A Name (SWAN)
- UNIAMO - Federazione Italiana Malattie Rare
- ALAPA - Alianza Argentina de Pacientes



www.iss.it/web/iss-en/rare-diseases

A *new web area* totally dedicated to Patients.

www.udninternational.org

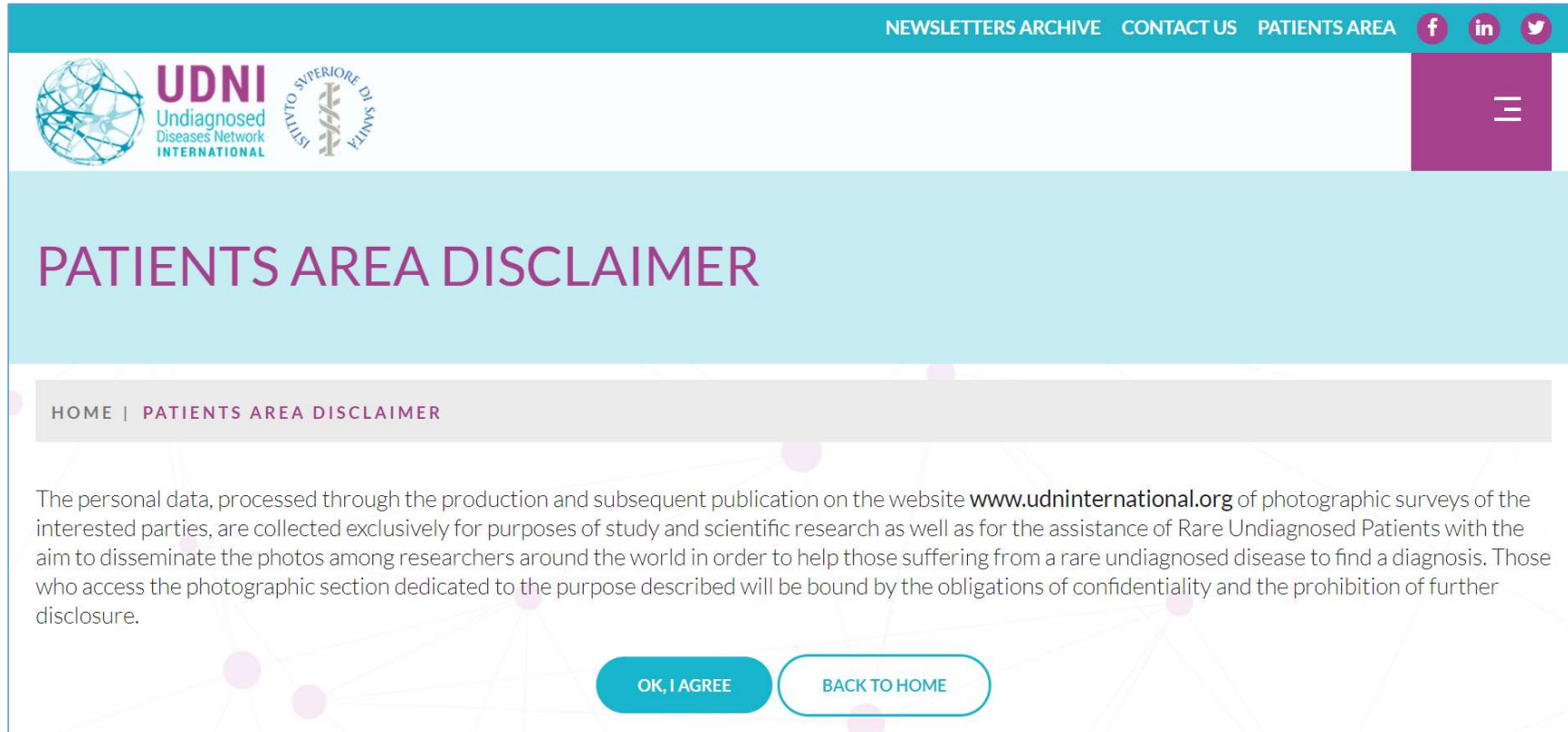
The screenshot shows the website's navigation bar with links for NEWSLETTERS ARCHIVE, CONTACT US, and PATIENTS AREA. The 'PATIENTS AREA' link is circled in green. Below the navigation bar is a banner for the 'UDNI CONFERENCE' featuring the '9th Conference of Undiagnosed Diseases Network International & Mayo Clinic Science Session' held from April 9-11, 2021. The banner includes a 'Call for Abstract, Registration and Draft Programme' and a 'MORE INFO' button. Logos for UDN International and the Istituto Superiore di Sanità are visible in the top left corner.



www.iss.it/web/iss-en/rare-diseases





This area is totally compliant with the European General Data Protection Regulation (**GDPR, art. 13 EU Regulation 2016/679**) and to its Privacy Code (as amended by European Legislative Decree 101/2018).



The screenshot shows the top navigation bar with links for NEWSLETTERS ARCHIVE, CONTACT US, and PATIENTS AREA, along with social media icons for Facebook, LinkedIn, and Twitter. The UDNI logo (Undiagnosed Diseases Network International) and the logo of the Istituto Superiore di Sanità are visible in the top left. The main heading is "PATIENTS AREA DISCLAIMER". Below the heading is a breadcrumb trail: HOME | PATIENTS AREA DISCLAIMER. The main text states: "The personal data, processed through the production and subsequent publication on the website www.udninternational.org of photographic surveys of the interested parties, are collected exclusively for purposes of study and scientific research as well as for the assistance of Rare Undiagnosed Patients with the aim to disseminate the photos among researchers around the world in order to help those suffering from a rare undiagnosed disease to find a diagnosis. Those who access the photographic section dedicated to the purpose described will be bound by the obligations of confidentiality and the prohibition of further disclosure." At the bottom of the text area are two buttons: "OK, I AGREE" and "BACK TO HOME".



NEWSLETTERS ARCHIVE CONTACT US PATIENTS AREA   

 **UDNI**
Undiagnosed
Diseases Network
INTERNATIONAL 

PATIENTS AREA

HOME | PATIENTS AREA

The UDNI together with the Wilhelm Foundation will offer undiagnosed patients (adults and children), who have been evaluated at an UDP and still are undiagnosed to share your story.

This is a tiny opportunity to a diagnosis. But if you still are undiagnosed don't hesitate to take this chance to show all the visitors your medical history and photos. Many of the specialists who visit the UDNI's website work all day long to solve the undiagnosed diseases and perhaps one of them find your child's story like one of the doctor's patient. If so, he will become interested and will contact you or your doctor at the UDP. We can't promise anything, but this is a new chance to a diagnosis.

This area is under the Wilhelm Foundation responsibility. In this section Wilhelm Foundation will collect some information (phenotypic, genotypic, picture) from undiagnosed rare patients.

For this purpose a dedicated ethics-Informed Assent as well as a UDNI Parental Consent were developed and approved during the 6th Conference of Undiagnosed Diseases Network International (19-21 June 2018, Naples) by all UDNI Board Members and participants. Before adding information on a patient these consents have to be download and signed by the Responsibles of patient.

Patients must have undergone a specific visit in a dedicated undiagnosed clinical site before being added in the present section.

ATTACHMENTS (new version with DPO consent to be used for UDNI purposes)

Ethic Informed Assent; Wilhelm Foundation Privacy; Informed consent for publication of patient images

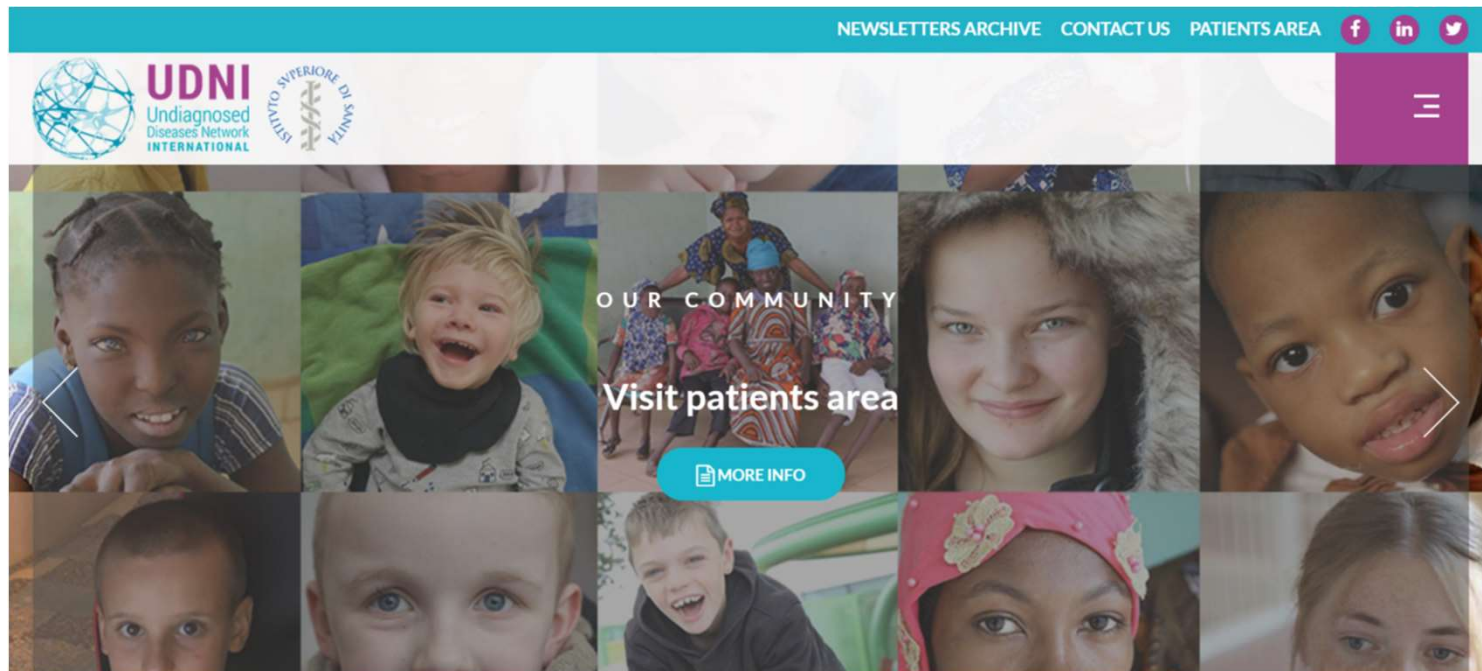


www.iss.it/web/iss-en/rare-diseases



Pictures of URD patients coming from Wilhelm Foundation are posted on the front page of the website with the aims to illustrate, to study, to improve knowledge and (hopefully) to reach a diagnosis to Patients with an unsolved condition.

https://www.udninternational.org/en-ricerca_patients



- **Forty-eight patients need for a definitive diagnosis.**
- **Images are rotating at least once a week to change their position in the website as well.**

<https://www.udninternational.org/schede-154-edwin>

HOME | PATIENTS AREA



Edwin is Undiagnosed

Date of Report

9

APR
2021

Edwin

Spasms in arms and legs, is very stiff. Involuntary movements, he can not sit or move. He is having difficult to grip things, harder with the left hand. He still has infant reflexes.

GENDER AND YEAR OF BIRTH

Male, 2019

DESCRIPTION

Edwin has spasms in arms and legs, is very stiff. Involuntary movements, he can not sit or move. He is having difficult to grip things, harder with the left hand. He still has infant reflexes. Has downs when he can't/want do anything. Afraid of sudden sounds. In periods he can't lie on his back. He can't turn from from his back to his stomach, can't keep his head up for a extended period of time when on his stomach.

He motor skills are on 5-6 months level. Intellectually around 1 year maybe. He does understand when spoken to. He doesn't have speech

A pseudo deficiency galactocerebrosidase

SYMPTOMS / SIGNS

- Spasms
- Tense
- Difficulties eating solid food
- Still has newborn reflexes
- Episodes when he can't/want do anything
- Motor difficulties
- Reflux
- Stomach problems
- Scream attacks
- Discomfort
- Smells like acetone
- Difficulty sleeping
- Apathetic
- Empty gaze
- Spikes of feve
- Perspiration
- Clenched fists

CURRENT TREATMENTS

- Clonidine
- Baclofen
- Nexium
- Melatonin
- Laxative
- Alimemazine

PRIOR TREATMENTS

- None

CONSIDERED TREATMENTS

- Baclofen pump

PREVIOUSLY CONSIDERED DIAGNOSES

- Krabbe disease, Cerebral Palsy

GENETIC VARIANTS OF INTEREST

- WGS Metabolic panel without findings

SIBLINGS, AGE & GENDER

Healthy Sisters 2015, 2018. Twinsister Ellie 2019 motorskills at 1 year level - she also understands when

CONTACT

CONTACT

If this participant sounds like you or someone you know, please contact Wilhelm Foundation at info@wilhelmfoundation.org.

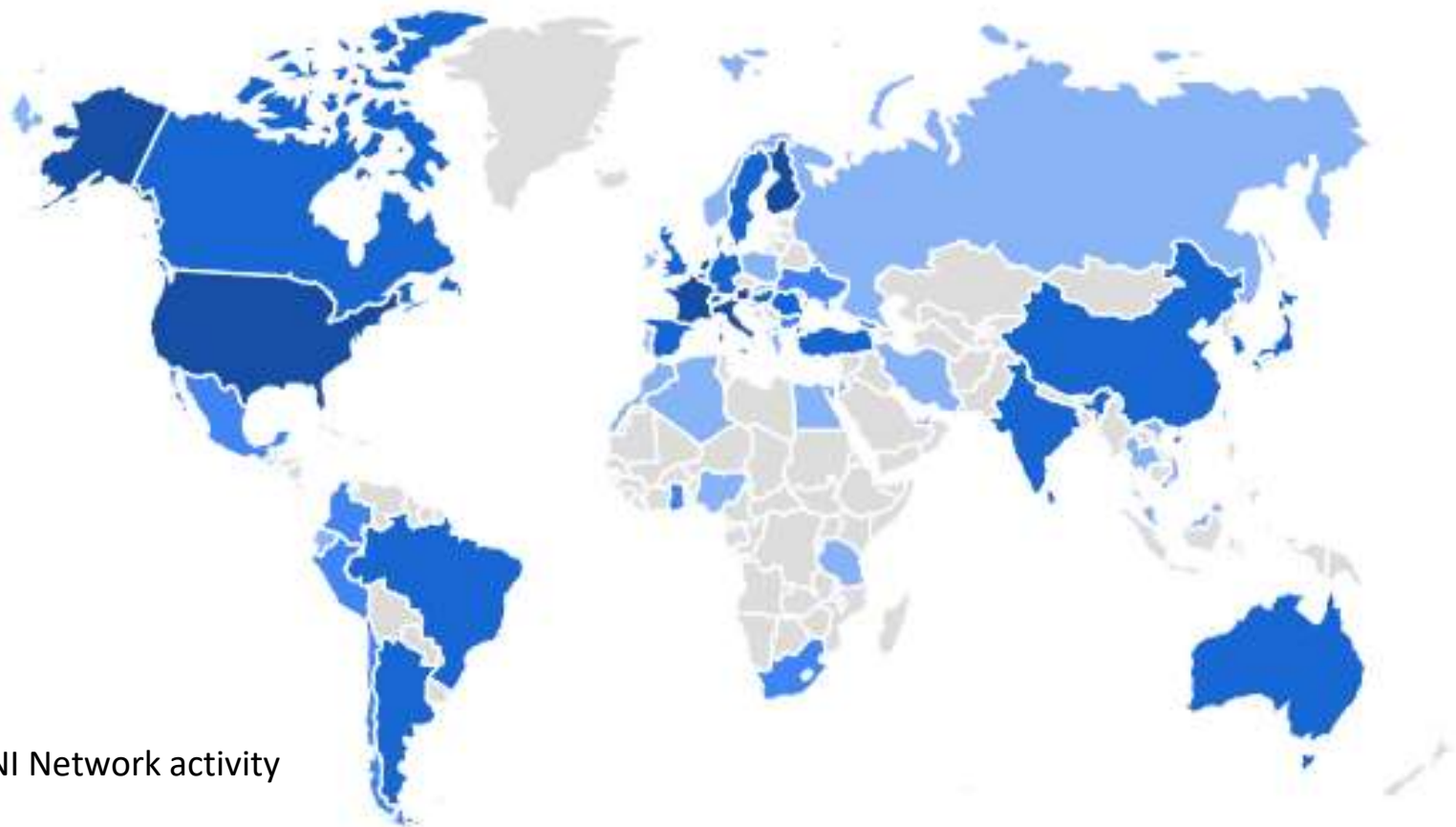
BIO

Edwin is boy who enjoys pranking and likes to play with his sisters. He loves when the include him in the play or drives him around. He loves to paint even though the paper is tasty to eat too. He likes to play with water and loves to bathe. He wants to do as much as he can and is very stubborn.

MORE PICTURES



www.iss.it/web/iss-en/rare-diseases



Global UDNI Network activity

Dark blu: very intense website activity



www.iss.it/web/iss-en/rare-diseases





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SELECT OUR SOCIAL!



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<https://twitter.com/UDNIss>

**UNDIAGNOSED DISEASE NETWORK
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STRATEGIC PARTNER OF MJC RUD**

**Working together:
in all UDNI activities
in the UDNI Working Groups
in other initiatives i.e.
International Courses**



THANK YOU
FOR YOUR ATTENTION



SEE YOU SOON...



Turin 10th UDNI conference, 31 January – 1 February, 2022



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