- NGO founded in Sweden in January 2009 Reg. No. 802444-3015
- Founding members FFDN and the Thalidomide Trust
- Aims:
  - Preserve the knowledge of the thalidomide survivors
  - Build an international European community
  - Launch and maintain an information website

EDRIC European Dysmelia Reference Information Centre

EDRIC Nijmegen 13th October 2017 michi.moik@edric.eu

European Federation

- 32 member organisations
- 18 countries
- 5 000 people represented
- 250 000 affected in EU
- 45 rare disease syndromes
- Congenital limb differences

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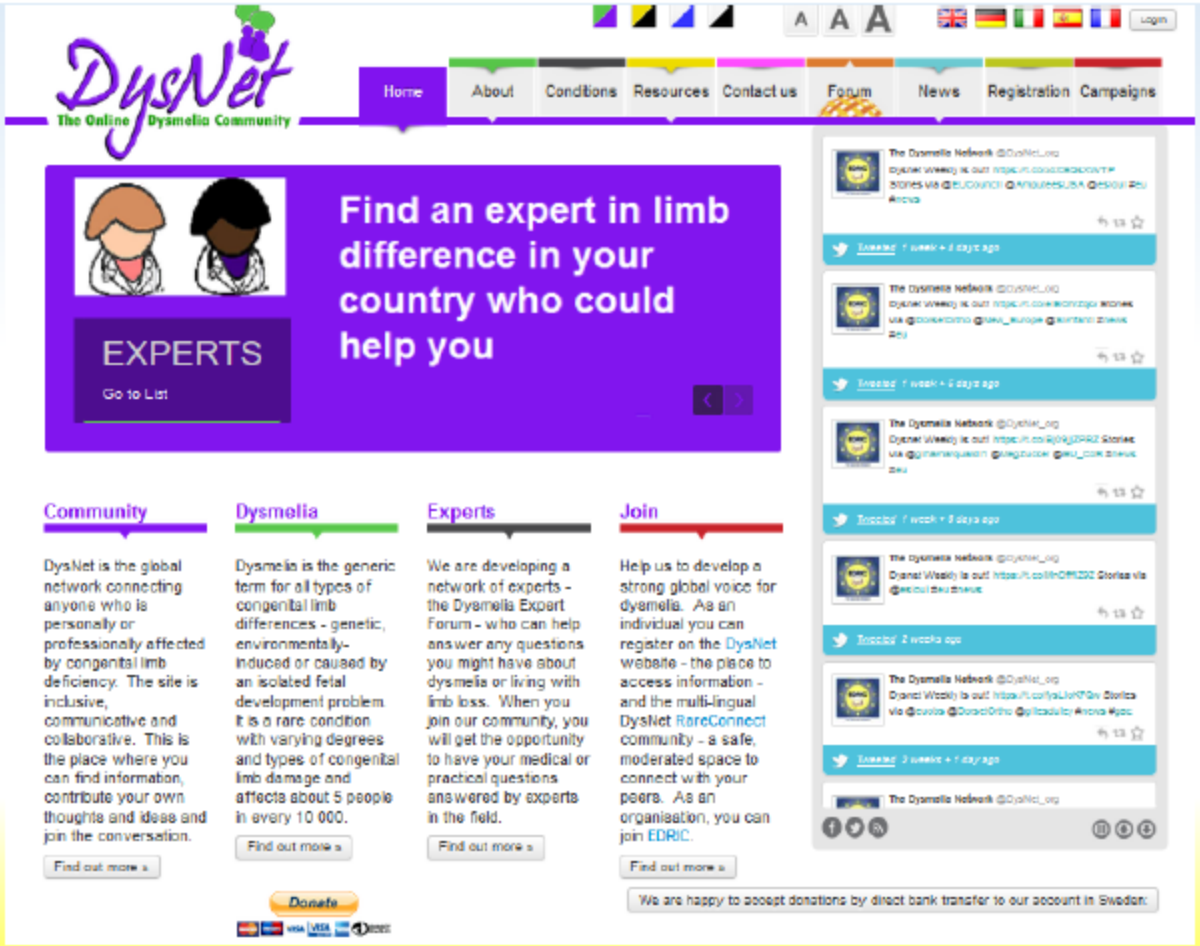
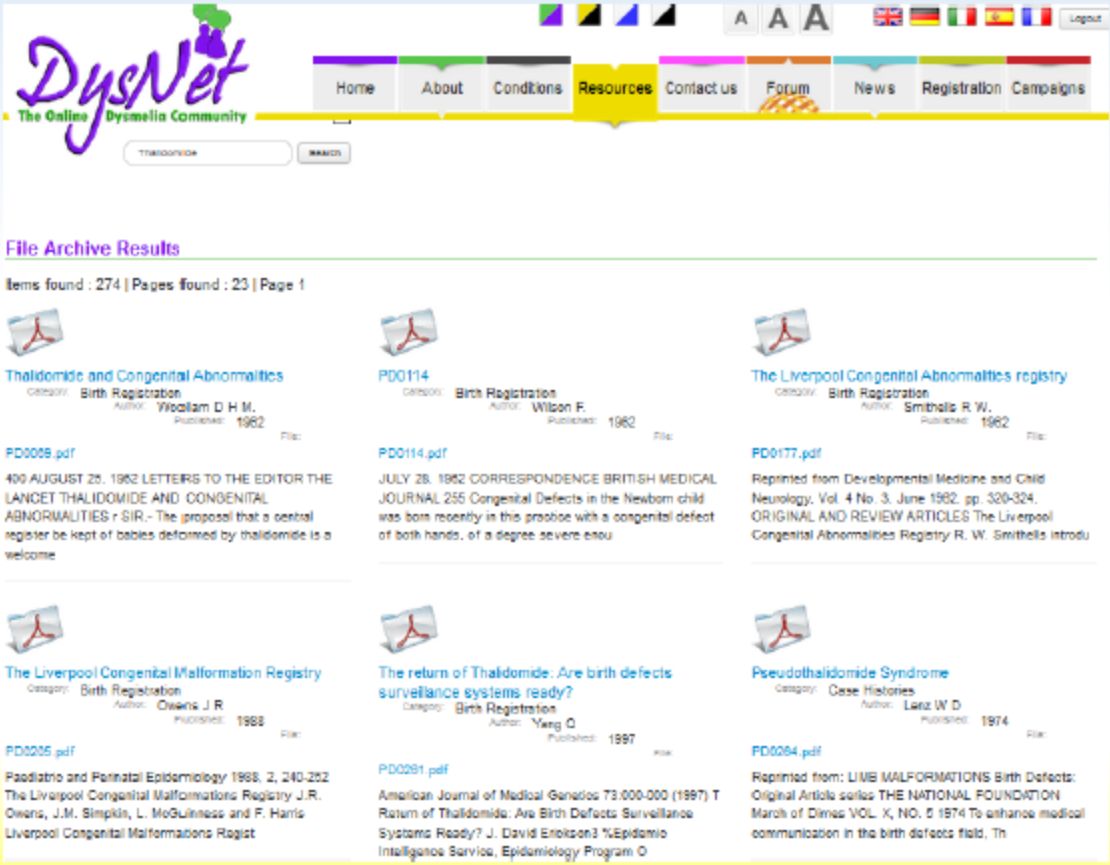
Dysnet.org

- Articles
- Professionals catalogue
- Forum
- Ressource database
- List of all conditions



Dysmelia Knowledge Base (DKB)

- Publication Database
- Eg for Thalidomide 274 entries



People with rare diseases/limb differences

- Geographical dispersion
- Health inequalities
- No highly specialised health services
  - Late diagnosis
  - Secondary conditions
  - Deterioration of health
  - Poor quality of life
  - Premature ageing

- 7-8% of the population has a rare condition

Issue Diagnosis

Study on 8 rare diseases Faurisson, F. (2007) found 40 % incorrect diagnoses

Average time to diagnosis 3,29 years with some diseases averages of 15 years, German action plan study (2009)Leads to

Late and incorrect health treatment malpractice

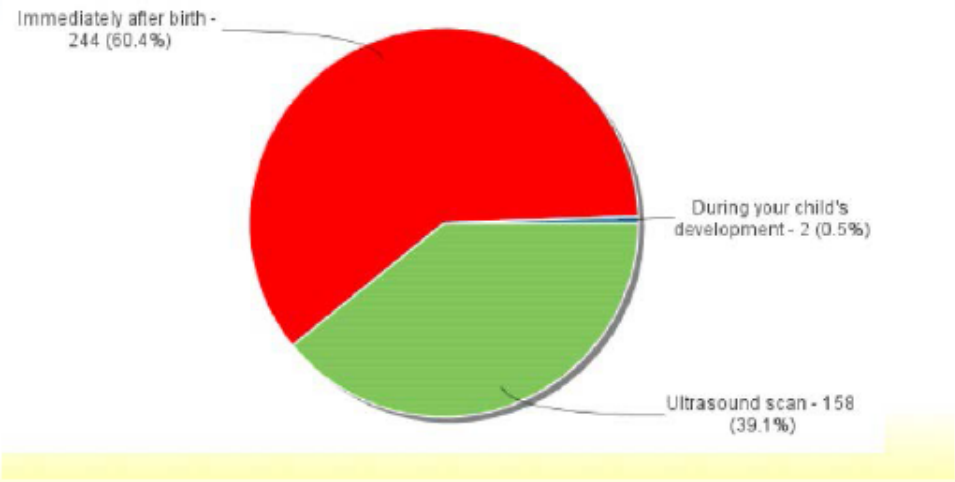
Gets even worse when sufficiently specialised health-services are not available

DysNet

What if?

Your Baby Has a Limb Difference

- EDRICs research with Univesity of Leeds. What-if your baby has a limb difference survey– Only 39,1 % of parents received the diagnosis at ultrasound.
- One of the reasons for this is in some countries like Sweden ultrasound examinations are not aimed at detecting limb differences



EDRIC TEAM

Charlotte Fielder, Geoff Adams-Spink, Tobias Arndt, Michi Moik, Björn Hakansson, Sal Giambruno, Monika Eisenberg-Geginat (v.l.n.r.)

Gemeinsam sind wir stark!

Dysmelien Syndrome

Humeroradial Synostosis,Radial Aplasia, 8q13 microdeletion syndrome, Adams-Oliver syndrome, Aglossia adactylia, Amelia, Amniotic band Syndrome, Apert Syndrome, Autosomal recessive amelia, Auyndrome, Brachydactyly, Campomelia; Cumming type, Cenani-Lenz syndrome, Ectrodactyly - split hand-foot malformation, SHFM, Fibula, Dimelia - diplopodia, Fibular hemimelia, Haas syndrome, Hanhart syndrome, Hemimelia, Holt-Oram syndrome, Microgastria, Microlissencephaly - micromelia oligodactyly, Phocomelia; Schinzel type, Poland syndrome, Polydactyly, Polymelia, Polysyndactyly, Radial hemimelia, Roberts syndrome (SC phocomelia), Sirenomelia, Symbrachydactyly, Syndactyly, Tetraamelia - multiple malformations, Tibial aplasia - ectrodactyly, Tibial hemimelia, Trisomy 18 syndrome, Ulbright-Hodes syndrome, Ulnar hemimelia, Thrombocytopenia-absent radius syndrome

EDRIC

DysNet

The Dysmelia Network

DysNet

Ideale, überall zugängliche Informationen

VON Menschen mit Dysmelie

FÜR Menschen mit Dysmelie

Wir bieten

- Informationen
- Erfahrungsaustausch
- Aktualität
- Fachwissen
- Netzwerk in 5 Sprachen
- Unterstützung
- Beratung

Spezielle Bedürfnisse erfordern spezielles Wissen und spezielle Behandlung

Menschen mit Dysmelie sind selten und stehen nicht im Fokus der Schulmedizin. Daher erfordern diese körperlichen Besonderheiten innovative (u.a. technische) Erleichterungen, Erfindergeist und Alternativen. Durch intensiven Ideen- und Erfahrungsaustausch von Menschen mit Dysmelie werden:

- neueste Erfahrungen gemacht und geteilt z.B. Hilfsmittel, Institutionen, Ärzte und Therapeuten
- umfassende Ressourcen geschaffen, welche online überall und jederzeit zugänglich sind.

Wo finde ich was im Internet

www.edric.eu

www.dysnet.org

Kontakt

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DYSNET
The Online Dysmelia Community
E-Mail: *info@dysnet.org*

Spendenkonto

Handelsbanken Schweden

IBAN: SE27 6000 0000 0004 3629

BIC: HANDSESS

Video

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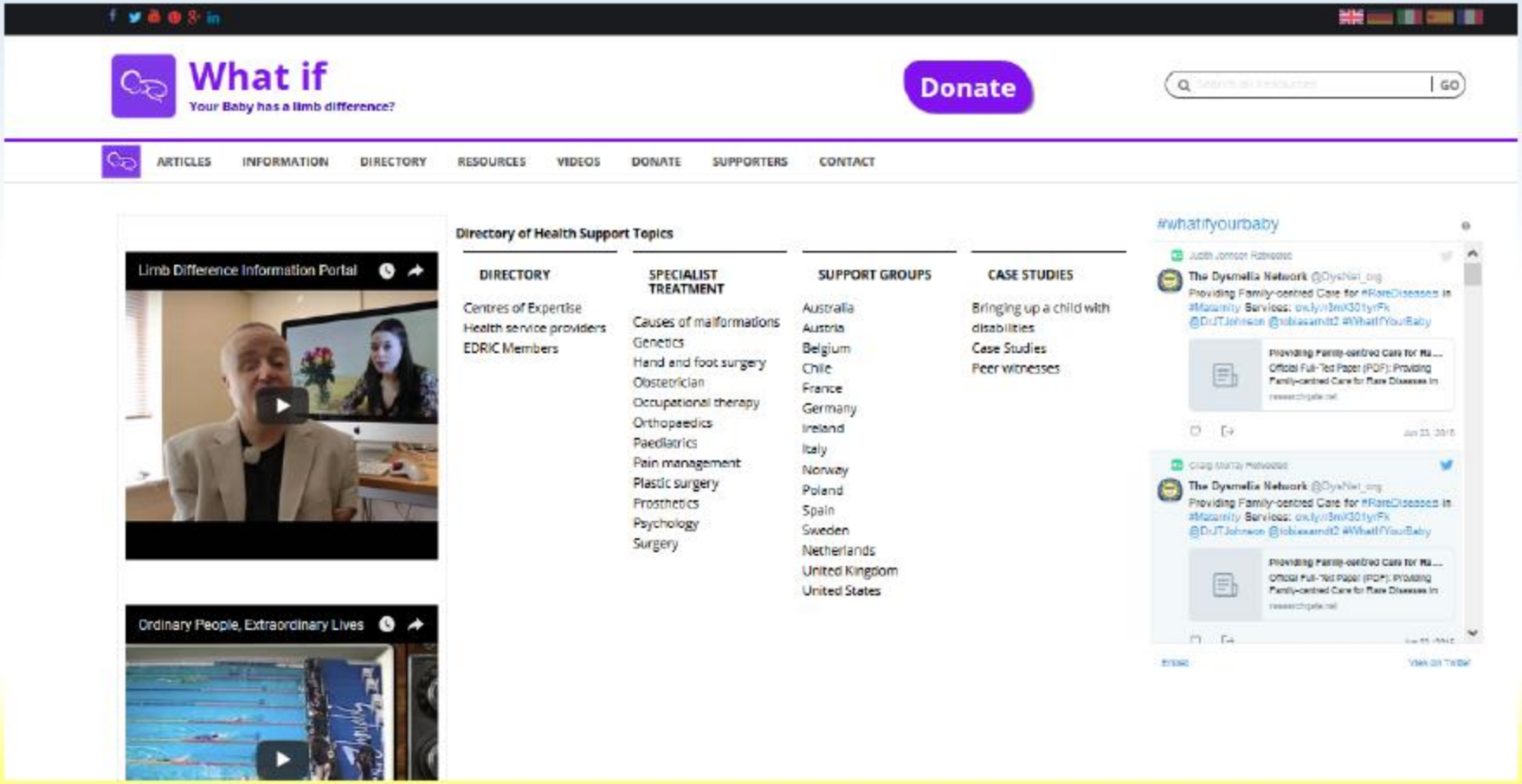

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Dysmelia Experts' Forum

- Stockholm October 8-11 2015
- Experts and EDRIC members come together in one forum
- 15 presenters, 90 participants
- 10 presentations
- 5 round-tables

Our dream: International Expert Network

Match patients and specialists

- Quality health information
- Direct support
- Including centres of expertise
- (e.g. Ex-Center)
- Enables cross-border healthcare
- Creates new health services
- Generates research

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EUROPEAN REFERENCE NETWORKS

FOR RARE, LOW PREVALENCE AND COMPLEX DISEASES

Share. Care. Cure.

European Reference Networks

The first 24 ERNs were launched in 2017, involving more than 900 highly-specialised healthcare units from over 300 hospitals in 26 Member States.

•Rare immunological and auto-inflammatory diseases•Rare bone diseases•Rare cancers* and tumours•Rare cardiac diseases•Rare connective tissue and musculoskeletal diseases•**Rare malformations and developmental anomalies and rare intellectual disabilities**•Rare endocrine diseases•Rare eye diseases•Rare gastrointestinal diseases•Rare gynaecological and obstetric diseases•Group on Cancer Control•Rare haematological diseases•Rare craniofacial anomalies and ENT (ear, nose and throat) disorders•Rare hepatic diseases•Rare hereditary metabolic disorders•Rare multi-systemic vascular diseases•Rare neurological diseases•Rare neuromuscular diseases•Rare pulmonary diseases•Rare renal diseases•Rare skin disorders•Rare urogenital diseases

ERN ITHACA (Intellectual disability TeleHealth And Congenital Anomalies)

- Main disease areas
- The network aims to cover rare intellectual disability and malformation syndromes,
- both diagnosed and undiagnosed,
- which require multidisciplinary approach to diagnosis and management.
- It is structured into four main thematic groups:

1. Rare congenital malformations not covered by other specific groups,
2. Malformation syndromes,
3. Rare intellectual disability, and
4. Rare chromosome disorders.

ERN ITHACA

Members

Coverage

No. HCP and Country – 33 HCPs across 12 EU countries

Austria	Belgium	Bulgaria	Croatia	Cyprus	Czech Republic	Denmark	Estonia	Finland	France	Germany	Greece	Hungary	Ireland	
					1			1	8	3		1		8
Latvia	**Liechtenstein**	**Lithuania**	**Luxemburg**	**Malta**	**Netherland**	**Norway**	**Poland**	**Portugal**	**Romania**	**Slovakia**	**Slovenia**	**Spain**	**Sweden**	
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*C – Coordinating Centre

ERN ITHACA

Added value

- Merging expertise and scattered patient cohorts under this network is one of the greatest benefits.
- Pooling of knowledge increases diagnosis and improves management
- Development of telemedicine initiatives and virtual multidisciplinary clinical review meeting and online clinics
- Long term follow-up review of cases and overseeing the transition from children to adulthoo
- Development of evidence based guidelines and natural history studies
- Development of patient registries
- Improvement of training in clinical trials and increase recruitment to clinical trial and studies
- Improvement of access to new diagnostic technologies – exome and genome sequencing – i the network and through research studies

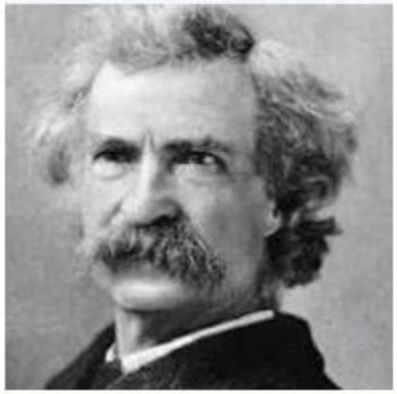
The network will provide and thus help our local doctors:

- ✓ Online training or webinars
- ✓ Focus training on new and emerging technologies and investigations
- ✓ Review of publications and journals
- ✓ Develop a curriculum for a Masters level degree in rare diseases working with Union Europeenne de Medicines Specialistes
- ✓ Coordinate training attachments and visits for trainees, senior clinicia and scientific staff and lay members to centres in the network.

Quality health information is key

Share and disseminate this information

Mark Twain: Be careful about reading health books. You might die of a misprint.



Dank U!

Michi Moik – Chairwoman EDRIC
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