



EDRIC European Dysmelia Reference Information Centre
EDRIC Nijmegen 13th October 2017 michi.moik@edric.eu

- 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



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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

DysNet
The Online Dysmelia Community

Home About Conditions Resources Contact us Forum News Registration Campaigns

Find an expert in limb difference in your country who could help you

EXPERTS
Go to List

Community
DysNet is the global network connecting anyone who is personally or professionally affected by congenital limb deficiency. The site is inclusive, communicative and collaborative. This is the place where you can find information, contribute your own thoughts and ideas and join the conversation.
[Find out more »](#)

Dysmelia
Dysmelia is the generic term for all types of congenital limb differences - genetic, environmentally-induced or caused by an isolated fetal development problem. It is a rare condition with varying degrees and types of congenital limb damage and affects about 5 people in every 10 000.
[Find out more »](#)

Experts
We are developing a network of experts - the Dysmelia Expert Forum - who can help answer any questions you might have about dysmelia or living with limb loss. When you join our community, you will get the opportunity to have your medical or practical questions answered by experts in the field.
[Find out more »](#)

Join
Help us to develop a strong global voice for dysmelia. As an individual you can register on the DysNet website - the place to access information - and the multi-lingual DysNet RareConnect community - a safe, moderated space to connect with your peers. As an organisation, you can join EDRIC.
[Find out more »](#)

[Donate](#)

We are happy to accept donations by direct bank transfer to our account in Sweden.



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Dysmelia Knowledge Base (DKB)

- Publication Database
- Eg for Thalidomide 274 entries

The screenshot shows the DysNet website interface. At the top, there is a navigation bar with links: Home, About, Conditions, Resources (highlighted), Contact us, Forum, News, Registration, and Campaigns. Below the navigation bar is a search bar with the text 'Thalidomide' and a 'SEARCH' button. The main content area is titled 'File Archive Results' and shows 'Items found : 274 | Pages found : 23 | Page 1'. The results are displayed in a grid format, showing the title, category, author, and publication year for each entry. The first entry is 'Thalidomide and Congenital Abnormalities' by Woolam D H M., published in 1962. The second entry is 'The Liverpool Congenital Abnormalities registry' by Smithells R W., published in 1962. The third entry is 'The return of Thalidomide: Are birth defects surveillance systems ready?' by Yang Q., published in 1997. The fourth entry is 'Pseudothalidomide Syndrome' by Lenz W D., published in 1974. The fifth entry is 'Paediatric and Perinatal Epidemiology 1968, 2, 240-252' by Owens, J.M., Simpkins, L., McGuinness and F. Harris, published in 1968. The sixth entry is 'American Journal of Medical Genetics 73:600-600 (1997) T Return of Thalidomide: Are Birth Defects Surveillance Systems Ready?' by J. David Erickson, published in 1997. The seventh entry is 'Reprinted from: LMB MALFORMATIONS Birth Defects: Original Article series THE NATIONAL FOUNDATION March of Dimes VOL. X, NO. 5 1974 To enhance medical communication in the birth defects field. Th' by R. W. Smithells, published in 1974.

Thalidomide and Congenital Abnormalities
Category: Birth Registration
Author: Woolam D H M.
Published: 1962
File: PD0009.pdf
400 AUGUST 25, 1962 LETTERS TO THE EDITOR THE LANCET THALIDOMIDE AND CONGENITAL ABNORMALITIES r SIR.- The proposal that a central register be kept of babies deformed by thalidomide is a welcome

The Liverpool Congenital Abnormalities registry
Category: Birth Registration
Author: Smithells R W.
Published: 1962
File: PD0177.pdf
Reprinted from Developmental Medicine and Child Neurology, Vol. 4 No. 3, June 1962, pp. 320-324. ORIGINAL AND REVIEW ARTICLES The Liverpool Congenital Abnormalities Registry R. W. Smithells introdu

The return of Thalidomide: Are birth defects surveillance systems ready?
Category: Birth Registration
Author: Yang Q.
Published: 1997
File: PD0291.pdf
American Journal of Medical Genetics 73:600-600 (1997) T Return of Thalidomide: Are Birth Defects Surveillance Systems Ready? J. David Erickson3 %Epidemiol Intelligence Service, Epidemiology Program O

Pseudothalidomide Syndrome
Category: Case Histories
Author: Lenz W D.
Published: 1974
File: PD0284.pdf
Reprinted from: LMB MALFORMATIONS Birth Defects: Original Article series THE NATIONAL FOUNDATION March of Dimes VOL. X, NO. 5 1974 To enhance medical communication in the birth defects field. Th



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



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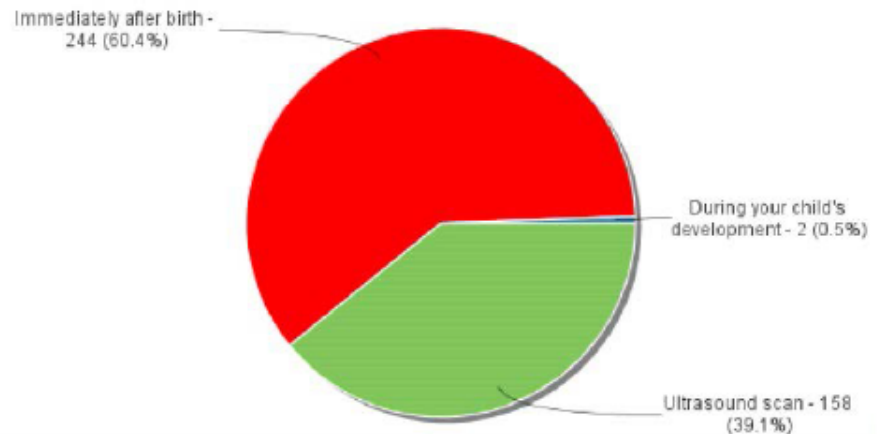
Your Baby Has a Limb Difference



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- EDRICs research with University 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

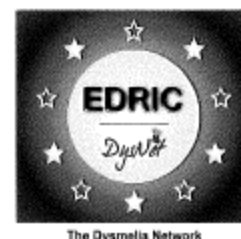


Charlotte Fielder, Geoff Adams-Spink, Tobias Arndt, Michi Moik, Björn Hakansson, Sal Giambruno, Monika Eisenberg-Geginat (v.l.n.r.)

*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



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

EDRIC

Axelstorpsvägen 2
Båstad
269 42
Sweden
E-Mail: info@edric.eu

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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What if

Your Baby has a limb difference?

Donate

Search all resources

GO



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CONTACT

Limb Difference Information Portal



Ordinary People, Extraordinary Lives



Directory of Health Support Topics

DIRECTORY

Centres of Expertise
Health service providers
EDRIC Members

SPECIALIST TREATMENT

Causes of malformations
Genetics
Hand and foot surgery
Obstetrician
Occupational therapy
Orthopaedics
Paediatrics
Pain management
Plastic surgery
Prosthetics
Psychology
Surgery

SUPPORT GROUPS

Australia
Austria
Belgium
Chile
France
Germany
Ireland
Italy
Norway
Poland
Spain
Sweden
Netherlands
United Kingdom
United States

CASE STUDIES

Bringing up a child with disabilities
Case Studies
Peer witnesses

#whatifyourbaby

Judith Johnson Retweeted

The Dystonia Network @DysNet.org
Providing Family-centred Care for #RareDiseases in
#Maternity Services: <https://doi.org/10.1186/s13059-017-1194-4>
@DrJTJohnson @robkassamdt2 #WhatIfYourBaby



Jun 23, 2017

Chris Murray Retweeted

The Dystonia Network @DysNet.org
Providing Family-centred Care for #RareDiseases in
#Maternity Services: <https://doi.org/10.1186/s13059-017-1194-4>
@DrJTJohnson @robkassamdt2 #WhatIfYourBaby



Jun 23, 2017

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View on Twitter



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Dysmelia Expert Forum Conference
Stockholm - October 8-11
2015



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





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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**





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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



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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.



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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	Italy
					1			1	8	3		1		8
Latvia	Liechtenstein	Lithuania	Luxembourg	Malta	Netherlands	Norway	Poland	Portugal	Romania	Slovakia	Slovenia	Spain	Sweden	Switzerland
		1			5			1	1				1	2

*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 meetings and online clinics
- Long term follow-up review of cases and overseeing the transition from children to adulthood
- 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 – in the network and through research studies



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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 clinicians and scientific staff and lay members to centres in the network.

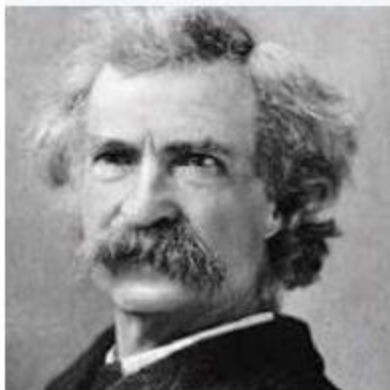


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Quality health information is key

Share and disseminate this information

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





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Dank U!

Michi Moik – Chairwoman EDRIC

EDRIC – European Dysmelia Reference Information Center

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