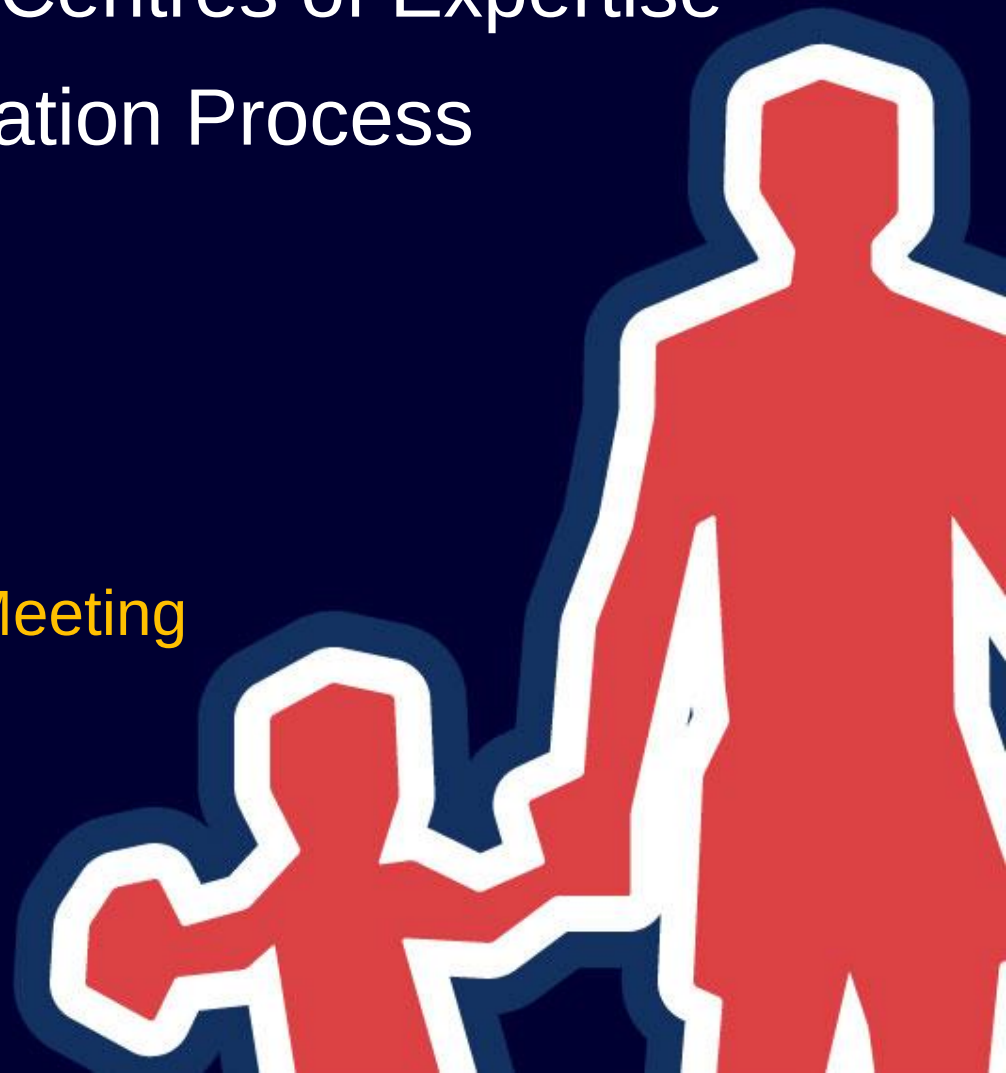
National Alliance for Rare and Genetic Diseases VSOP
- The Netherlands -

VSOP

Connecting Patient Organisations to Centres of Expertise by a Legal Role in the Designation Process

Dr. Cor Oosterwijk
Director VSOP

EURORDIS Membership Meeting
14 May 2021



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VSOP has 90+ members

ADCA Vereniging Nederland • ALS Patients Connected (APC) • Amyloïdose Nederland • Bardet-Biedl Syndroom Stichting • Belangengroep MEN • Belangenvereniging LOA/LHON • Belangenvereniging van Kleine Mensen (BVKM) • Belangenvereniging Von Hippel-Lindau (VHL) • Care4BrittleBones • Christianson Syndrome Europe • CMTC-OVM Vereniging • Contactgroep Marfan Nederland • DEBRA Nederland • DSDNederland • FOP Stichting Nederland • Fragiele X Vereniging Nederland • Fabry Support & Informatie Groep Nederland (FSIGN) • Galactosemie Vereniging Nederland (GVN) • HEVAS • HME-MO Vereniging Nederland • Huidpatiënten Nederland • Vereniging voor Ichtyosis Netwerken • Interstitiële Cystitis Patiëntenvereniging (ICP) • Jeugdreuma Vereniging Nederland • Kans voor PKAN kinderen • KortMaarKrachtig vereniging voor geamputeerden (KMK) • Laposa: Landelijke Patiënten- en Oudervereniging voor Schedel- en/of Aangezichtsafwijkingen • LGD Alliance Nederland • Lichen Planus Vereniging • Longfibrose Patiëntenvereniging • Macula Vereniging • Mastocytose vereniging Nederland • MSS Research Foundation • Nederlandse Cystic Fibrosis Stichting (NCFS) • Nederlandse Hypofyse Stichting • Nederlandse Klinefelter Vereniging (NKV) • Nederlandse PKU Vereniging • Nederlandse Rett Syndroom Vereniging (NRSV) • Nederlandse Vereniging van Hemofilie-Patiënten (NVHP) • Nederlandse Vereniging voor Patiënten met Paragangliomen • NephcEurope • NFVN: Neurofibromatose Vereniging Nederland • NVN: Nierpatiënten Vereniging Nederland • NINA Foundation • OSCAR Nederland • Parkinson Vereniging • Patiëntenvereniging Fibreuze Dysplasie • Patiëntenvereniging MED SED • PCD Belangengroep • PlatformCHD • Prader-Willi Fonds • SAS: Stichting voor Afweerstoornissen • SCCH Vereniging • SGA Platform SOSNL • Spierziekten Nederland • Stichting AA & PNH Contactgroep • Stichting Cure ADOA • Stichting De Negende Van… • Stichting De Ontbrekende Schakel • Stichting Diagnose Kanker (SDK) • Stichting Downsyndroom (SDS) • Stichting GNAO1 • Stichting Lynch Polyposis • Stichting MRK-vrouwen • Stichting Noonan Syndroom • Stichting Overdruksyndroom NL • (SOSNL) • Stichting PHA Nederland • Stichting Pierre Robin Europe • Stichting RPF • Stichting Prader-Willi Syndroom • Stichting Rubinstein Taybi Syndroom • STSN: Stichting Tubereuze Sclerosis Nederland • Stichting IJzersterk • Patiëntenplatform Sarcomen • Syringomyelie Patiënten Vereniging • Vasculitis Stichting • Vereniging Anusatresie (VA) • Vereniging Cornelia de Lange Syndroom • Vereniging Oog in Oog • Vereniging van Allergie Patiënten (VAP) • Vereniging van Ehlers-Danlos patiënten (VED) • Vereniging van Huntington • Vereniging van patiënten met Erythropoëtische protoporphyrie • Vereniging voor Angio Oedeem • Vereniging ziekte van Hirschsprung • VKS: Volwassenen, Kinderen en Stofwisselingsziekten • VOI: Vereniging Osteogenesis Imperfecta • VOKS: Vereniging voor Ouderen en Kinderen met een Slokdarmafsluiting • ZeldSamen. Kandidates: Stichting Steun 22Q11 • Stichting Orthostatische Tremor • TAPS-support • XLH Vereniging Nederland • Vasa Previa Stichting

From Council Recommendation to National Plans

2009 2010 2011 2012 2013 2014 2015

European Commission instructs Member States to develop a National Plan for Rare Diseases

Dutch Nationaal Plan Rare Diseases

Dutch ministry of health gives assignment to set up an assessment procedure

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I. Patient involvement in the accreditation procedure

II. Connecting CoE’s

III. Connecting CoE’s with ERN’s

EUCERD criteria for designation of CoEs for RD in MS

1. Capacity to produce and adhere to good practice guidelines for diagnosis and care.
2. Quality management in place to assure quality of care, including National and European legal provisions, and participation in internal and external quality schemes when applicable.
3. Capacity to propose quality of care indicators in their area and implement outcome measures including patient satisfaction.
4. High level of expertise and experience documented, for instance, by the annual volume of referrals and second opinions, and through peer-reviewed publications, grants, positions, teaching and training activities.
5. Appropriate capacity to manage RD patients and provide expert advice.
6. Contribution to state-of-the-art research.
7. Capacity to participate in data collection for clinical research and public health purposes.
8. Capacity to participate in clinical trials, if applicable.
9. Demonstration of a multi-disciplinary approach, when appropriate, integrating medical, paramedical, psychological and social needs (e.g. RD board).
10. Organisation of collaborations to assure the continuity of care between childhood, adolescence and adulthood, if relevant.
11. Organisation of collaborations to assure the continuity of care between all stages of the disease.
12. Links and collaboration with other CoE at national, European and international level.
13. Links and collaboration with patient organisations where they exist.
14. Appropriate arrangements for referrals within individual Member States and from/to other EU countries if applicable.
15. Appropriate arrangements to improve the delivery of care and especially to shorten the time taken to reach a diagnosis.
16. Consideration of E-Health solutions (e.g. shared case management systems, expert systems for tele-expertise and shared repository of cases).

Governmental policy since 2021

- Legal national basis for the acrreditation of CoE, based upon the 2009 *'Council recommendation on a action the field of rare diseases'*
- Involvement of patient organisations and VSOP as a rule
- Connection of CoE with ERN: obligatory
- Several requirements for CoE's now with obligatory indicators

Announcements:

- Shortening diagnostic delay
- Concentration of CoE bases on national and ERN volume-standards
- Use of Orphanet nomenclature in electronic health records
- Reimbursement of eHealth consultations

https://zoek.officielebekendmakingen.nl/stcrt-2021-1924.html

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National Requirements and Indicators

12 Requirements specified by 27 Indicators, of which 15 obligatory (18 per 2023):

<u>The care pathway…</u>

1. Must be renewed each 5 year
2. Includes diagnostics, patient visits and follow-up
3. Describes the relationship with multidisciplinary team, and with the case manager
4. Describes the composition of the multidisciplinary team and the role of its members

<u>The multidisciplinary team…</u>

5. Its functioning is guaranteed for the next 5 years by education and training
6. Its functioning is guaranteed by succession of departing members
7. Has at least two medical specialists with specific knowledge of the rare disease
8. The CoE has all necessary medicinal disciplines in the hospital

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National Requirements and Indicators

Others:

9. The provision of care meets formal (inter)national standards

The CoE…

10. uses patient's experiences for improvement of care
11. is 24/7 accessible for patients and care-givers
12. is available for consultation
13. is actively involved in education of patients and care-givers
14. has a scientific track record
15. has at least one specific publication in case of an ultra-rare disease

Added per 2013:

16. Each new CoE has added value compared to the already existing CoE
17. The CoE sees sufficient patient numbers
18. The CoE contributes actively to its ERN

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CoE's in the Netherlands

- 340 existing CoE's

2021

- 322 (re)applications:
  204 existing 41 mergers, 77 new
- 983 unique orpha codes

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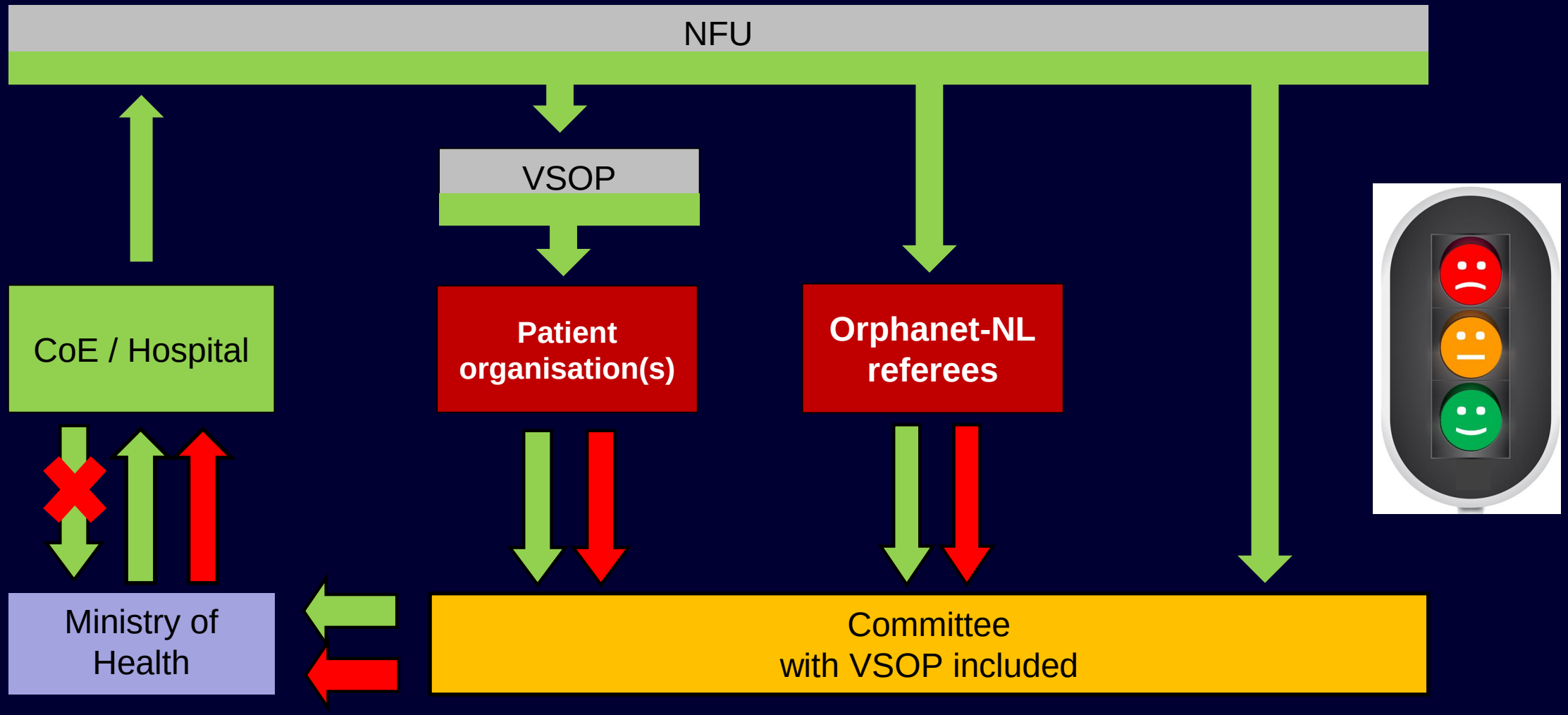
Accreditation Procedure



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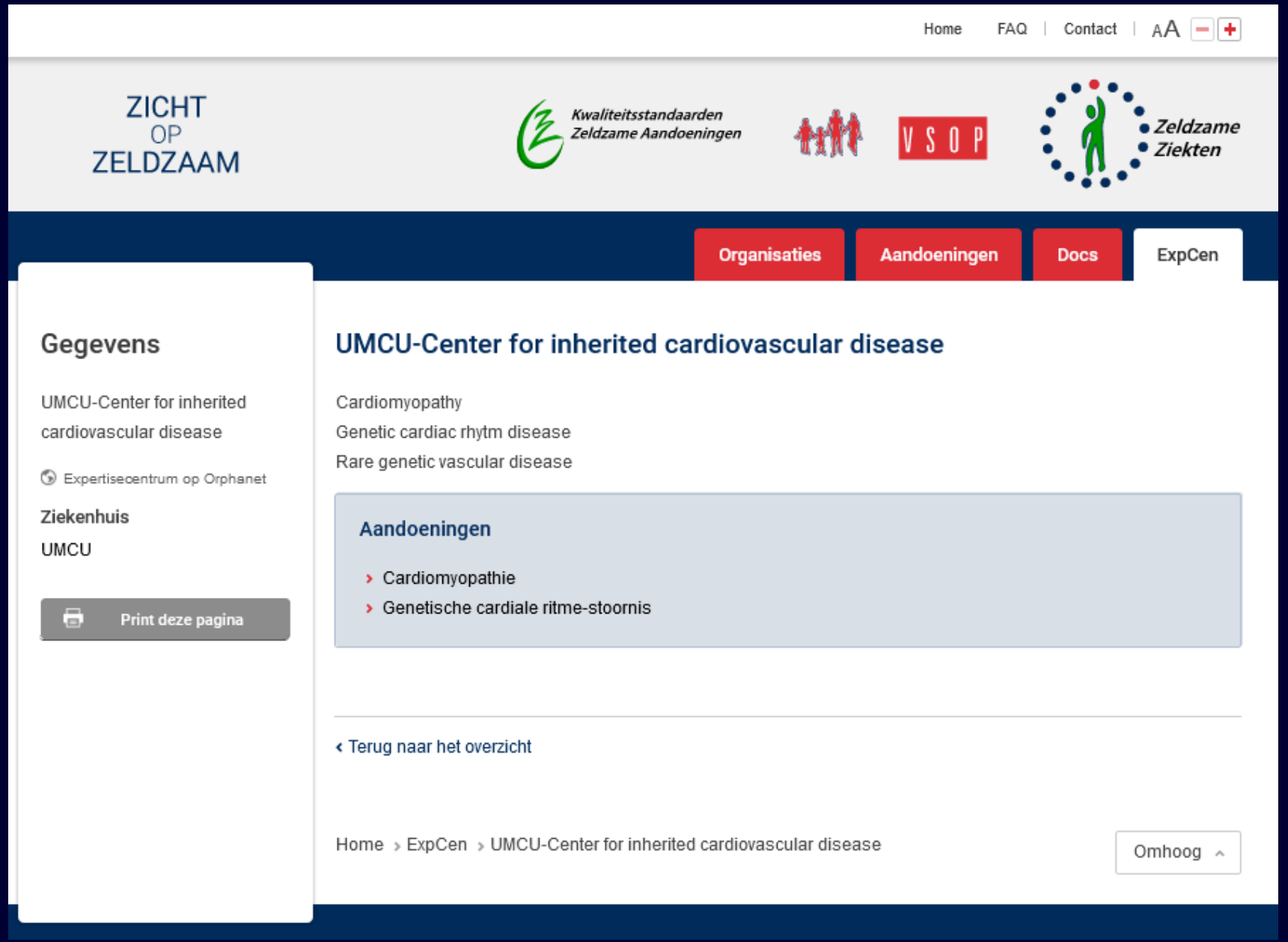
www.zichtopzeldzaam.nl



Patient involvement in the accreditation procedure - WHY ?

1) To prevent ‘self-accreditation’
2) To check EUCERD criterion # 13:

   “Links and collaboration with patient organisations where they exist.”
3) To stimulate cooperation between patient organizations and CoE’s
4) To guarantee the provision of holistic care from the patients’ perspective
5) To stimulate patient involvement in research
6) To stimulate referral of patients to the CoE by the patient organisation

➢ Suboptimally functioning CoE’s = Suboptimally functioning ERN’s !

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Connecting patient organisations to CoE's

- 190 (out of 280) participating patient organisations
- 1900 unique combinations of CoE – Orpha code – Patient organisation

Different CoE – same Orpha code

Nr EC	Aandoening	Orphacode	Patiëntenorganisatie
G-11-31	Carcinoma of the ampulla of Vater	ORPHA:300557	Alvleeskliervereniging (AVKV)
G-11-9	Carcinoma of the ampulla of Vater	ORPHA:300557	Alvleeskliervereniging (AVKV)
G-11-23	Carcinoma of the ampulla of Vater	ORPHA:300557	Alvleeskliervereniging (AVKV)

One CoE – two patient organisations

Nr EC	Aandoening	Orphacode	Patiëntenorganisatie
G-11-40	Soft tissue sarcoma	ORPHA:3394	ST Patiëntenplatform Sarcomen
G-11-40	Soft tissue sarcoma	ORPHA:3394	Vereniging Kinderkanker NL

Patient involvement in the accreditation procedure - HOW ?

VSOP asks the board of each patient organisation to confirm participation, and to check their rare diseases (orpha-codes)

VSOP offers the board a questionnaire

- 15 questions concerning the CoE
- 20 questions concerning each rare disease
- Final question: recognition of the CoE: YES or NO ?
- Strong points and points for improvement are forwarded to the CoE

VSOP offers a survey amongst their membership

- 17 questions
- board uses the outcome of this survey for their assessment

https://vsop.nl/wat-doen-wij/expertisecentra/beoordeling-expertisecentra/rol-patientenorganisaties/

Nice work. But what can I do in my country??

<u>The Dutch model could be applied in each country</u>

1) Agree upon a accreditation-procedure with your national Orphanet partner
2) Invite national hospital associations to participate
3) Lobby together for funding
4) Invite your government to participate

➢ Invite the Dutch Ministry of Health, and/or NFU, and/or VSOP for a visit

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Update National Plan in 2017

2013

2017

“ Everyone with a rare disease receives a timely, accurate diagnosis, shared care and treatment fitting one’s personal needs (cross border when needed).

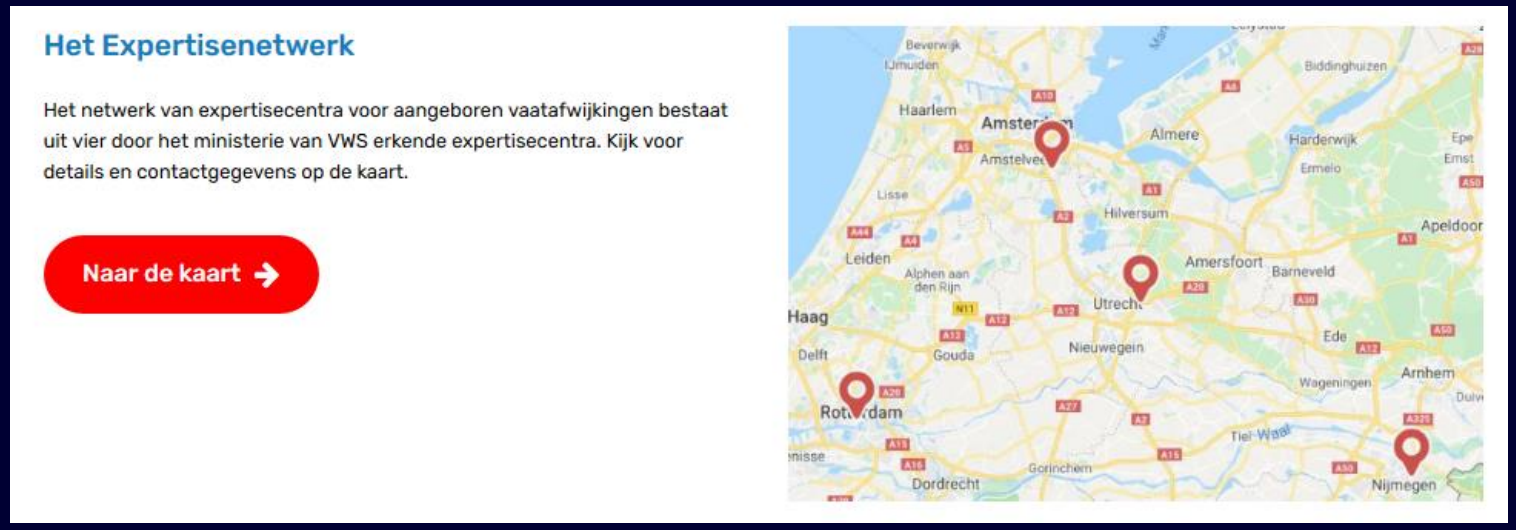
To make this possible, CoE’s needs to be able to fulfil all requirements, like creating networks with care-givers in the region where the patient is living, and with municipalities and health insurers.

Participation of people living with a rare disease and their organisations is a indissoluble part of this. “

National Networks of Expertise

"Further cooperation of CoE's within networks is needed for knowledge generation, and for patients and caregivers finding their way."

Dutch Minister of Health, 8 June 2018



"E-health consultations between CoE's and care-givers at other locations, must be reimbursed."

Dutch Minister of Health, 6 January 2021

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Networks of Expertise

1) https://mastocytose-expertise.net
2) https://aangeborenvaatafwijkingen-expertise.net
3) https://itp-expertise.net
4) https://nf1-expertise.net
5) https://amyloidose-expertise.net
6) https://arm-expertise.net
7) https://chd-expertise.net
8) https://slokdarmafsluiting-expertise.net
9) https://hirschsprung-expertise.net
10) https://psc-expertise.net

- 8 others under construction
- 11 neuromuscular networks: https://www.spierziektencentrum.nl/patientenzorg

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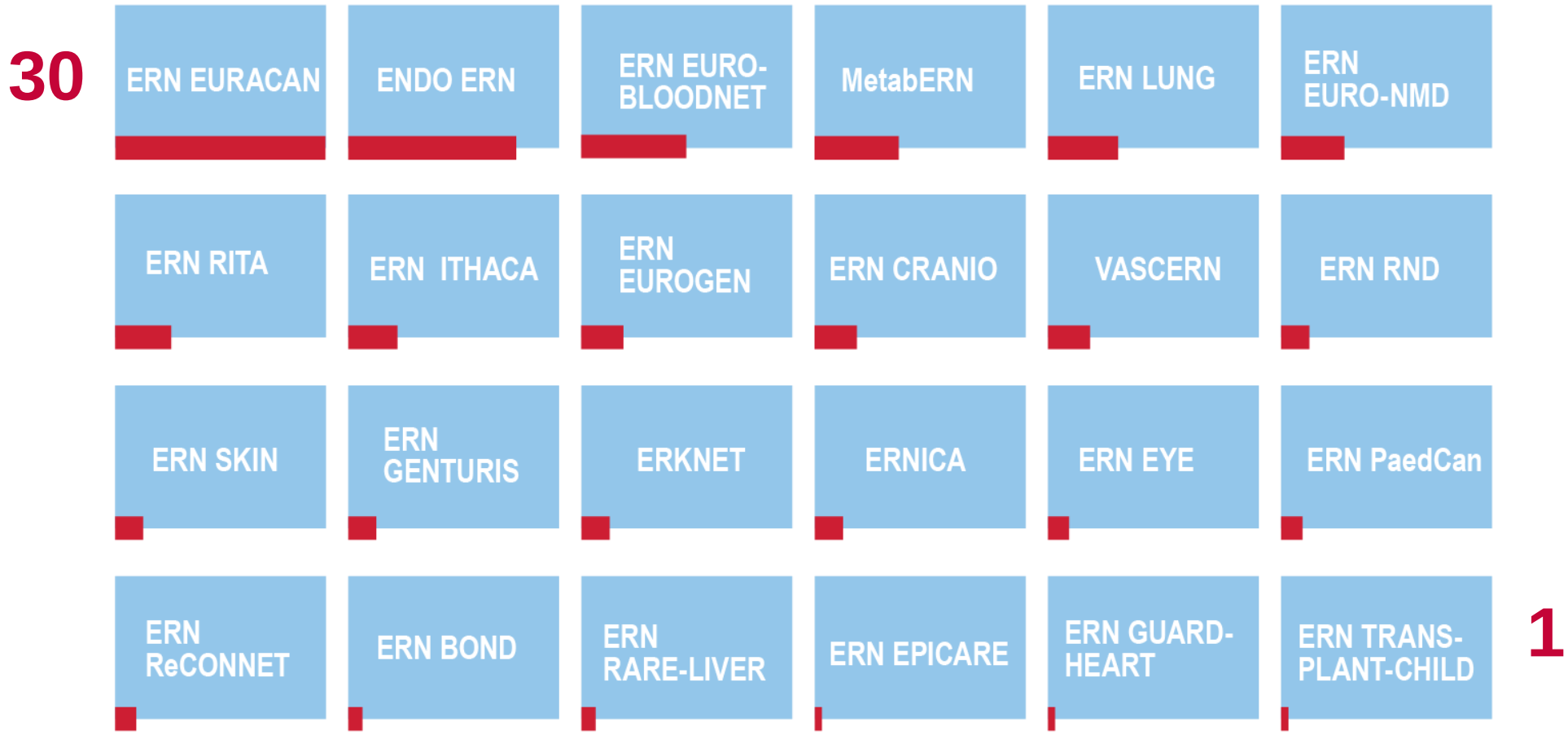
Participation of Dutch CoE's in ERN's

- 164 (+37 applications) out of 340 participating CoE's
- 7 ERN coordinators



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How to connect 300+ CoE's with 24 ERN's?

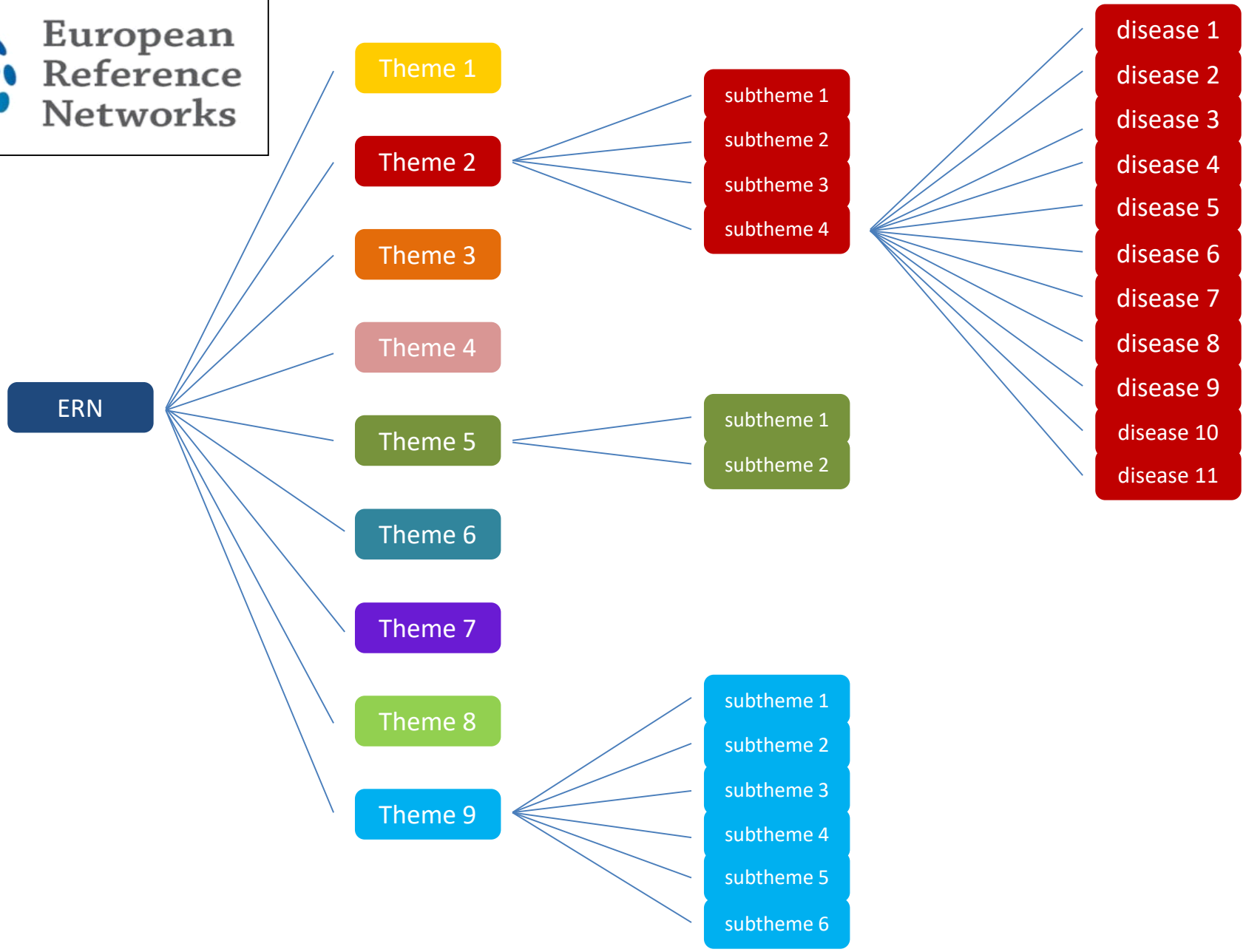
National networks could be based upon:

- Orpha-codes as specific as possible?
- Orpha-code groupings.
- ERNs (sub)themes?
- Hospitals?

<u>First steps</u>

- ➢ merging CoE's within hospitals, based on orpha-code groupings
- ➢ (Re)applications: accounting for ERN (sub)themes

European Reference Networks



NFU

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Analysis of ERN's classifications

	Information provided by ERN		Informatie Orphanet database
Network Code	Thematic area	Sub-thematic area	Orphanet data(Pat Id - naam groep/aandoening - [Typology] - ORPHAcode)
EURO-NMD	Muscle Diseases	Aquired skeletal muscle diseases	18538 Acquired skeletal muscle disease [Group of disorders - Category] ORPHA:206638
		Genetic skeletal	18537 Genetic skeletal muscle disease [Group of disorders - Category] ORPHA:206634
EURO-NMD	Neuromuscular Junction (NMJ) Defects	Myasthenia Gravis Other autoimmune NMJ defects (Lambert-Eaton)	24050 Immune-mediated acquired neuromuscular junction disease [Group of disorders - Clinical group] ORPHA:464764
		Congenital Myasthenic Syndromes	8737 Congenital myasthenic syndrome [Disorder - Disease] ORPHA:590
EURO-NMD	Peripheral Neuropathies	Inherited Neuropathies	13514 Genetic peripheral neuropathy [Group of disorders - Category] ORPHA:98497
		Acquired Neuropathies	18209 Acquired peripheral neuropathy [Group of disorders - Category] ORPHA:182086
EURO-NMD	Motor Neuron Disorders	Amyotrophic Lateral Sclerosis (ALS) and related diseases	106 Amyotrophic lateral sclerosis [Disorder - Disease] ORPHA:803
		Spinal Muscular Atrophy (SMA) and related diseases	633 Proximal spinal muscular atrophy [Disorder - Disease] ORPHA:70 / 10744 Distal hereditary motor neuropathy [Group of disorders - Clinical group] ORPHA:53739 / 18547 Bulbospinal muscular atrophy [Group of disorders - Category] ORPHA:206701
EURO-NMD	Mitochondrial diseases	Mitochondrial encephalomyopathies caused by mtDNA	10520 Mitochondrial disease [Group of disorders - Category] ORPHA:68380
		Nuclear gene defects causing mitochondrial encephalomyopathy	

‘Weten waar ik terecht kan voor de juiste zorg is zoveel waard’

JOUW ERVARING IS WAARDEVOL

Meld je aan voor het patiëntenpanel ZeldzaamInZicht

Visit our websites

- Corporate: vsop.nl
- Rare diseases: zeldzameaandoening.nl
- Quality of care: kwaliteitvoorzeldzaam.nl
- CoE’s: zichtopzeldzaam.nl
- Patient panel: zeldzaaminzicht.nl
- Registries: patientenregisters.org
- Pediatric research: kindenonderzoek.nl
- Syndromes: syndromen.net
- Diagnostic centres: diagnoseonbekend.nl
- No diagnosis: ziekteonbekend.nl
- Preconception care: preparingforlife.net

‘Diagnose zou een stuk sneller kunnen’

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