The organization of the management of rare diseases in Europe and France

A Verloes

European Commission

EUROPEAN REFERENCE NETWORKS

Helping patients with rare or low-prevalence complex diseases

maladies rares

European Reference Networks ITHACA

Rare diseases in Europe

OVER
6000
distinct rare diseases

Each one affects fewer than
1 IN
2000
PEOPLE

All together, an estimated
30
MILLION PEOPLE
are living with a rare disease in Europe

Expertise, knowledge, information on diseases and their consequences are **scarce and difficult to access**

Rare, complex, chronic, disabling, progressive, degenerative, often life-threatening

NO
CURE
for the vast majority of diseases and few treatments available

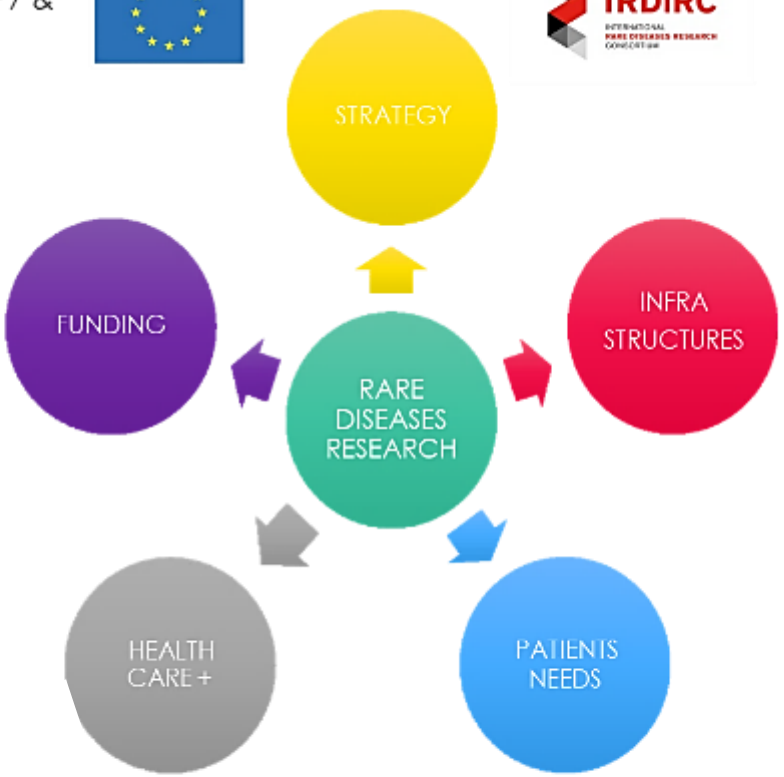
They are **geographically scattered** and often **isolated**

Few experts, geographically **scattered**

Research is fragmented

The complexity of rare disease landscape in EU

1.4 billion € for RD research in FP7 & Horizon 2020



IRDiRC

RD Connect ERDRI orphanet Joint Research Centre JRC SolveRD

ECRIN EUROPEAN CLINICAL RESEARCH INFRASTRUCTURE NETWORK eatris BBMRI-ERIC elixir EU-OPENSCREEN Chemical keys for life's locks ePTRI INFRAFRONTIER mouse disease models conect 4children

ERDERA

EJP RD

European Reference Networks

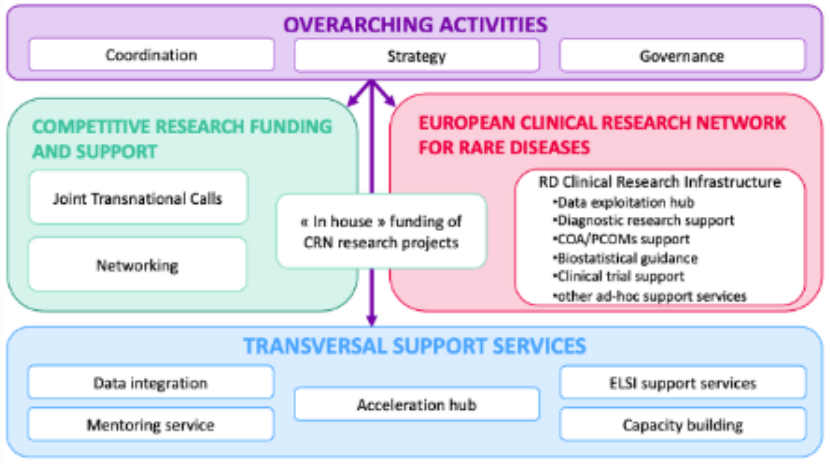
EURORDIS RARE DISEASES EUROPE

869 patient organisations from 76 countries

European Commission

Two EU initiative: care of RD (ERNs) & research on RD (ERDERA)

European Rare Diseases Research Alliance



The RD Partnership implementation concept

- **EJPRD** (European Joint Program on Rare Diseases)
  - EJPRD : Part of the EU H2020 Work Programme (budget of €55 M for 5 years) (2019-2024)
  - Consortium of 85 partners (29 research funding bodies/ministries and 5 charities/foundations, 12 research institutes + 33 universities & hospitals, 5 EU research infrastructures, EURORDIS & ePAGs

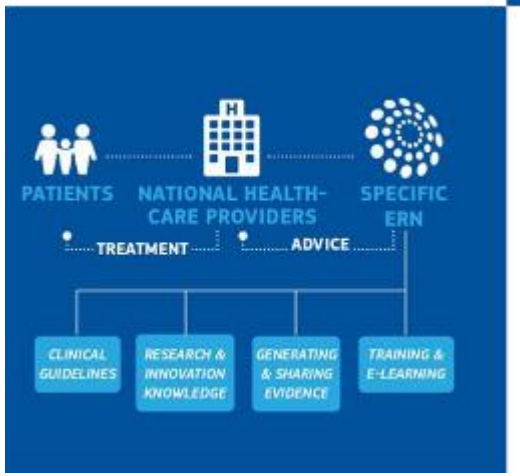
Replaced by **ERDERA** (under revision) : 2024-2031 - >150 M€

- ERDERA's objectives
  - **Improve the health** and well-being of the 30 million people in EU living with a rare disease (RD).
  - Support the **prevention, diagnosis and treatment** of rare diseases.
  - Commitment to the UN Sustainable Development Goals & Alignment with EU Policy Priorities
- ERDERA's Vision
  - **Structuring** the European Research Area on RD
  - Leave no one behind by establishing a **multi-stakeholder ecosystem** for RD
  - Supporting **patient-driven research**, developing new treatments and diagnostic pathways
  - Using Health and Research **Data** for Digital Transformational Change in RD R&I
  - **Coordinate and align national and regional research strategies**.
  - Establishing **public-private** collaborations and maximising the EU's innovation potential in R&D.
- ERDERA's missions
  - Gather and share R&I knowledge, resources and services for RD research projects.
  - Enable each consenting patient to participate in tailored **clinical studies** by generating and sharing regulatory-grade data.
  - Make Europe a world leader in RD R&I by increasing investment to boost innovation.

ERNs

- **European Reference Networks**

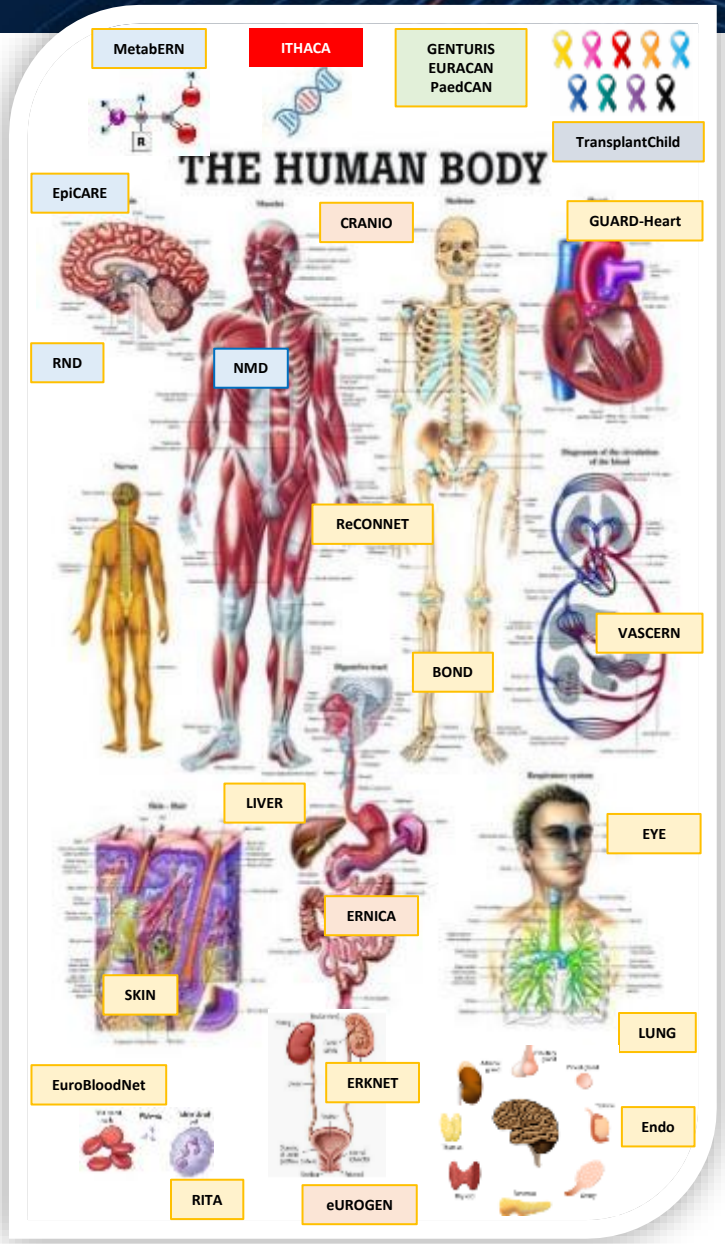
- Origine
  - Directive 2011/24/UE : « *droits des patients en matière de soins de santé transfrontaliers* »
- Objectives
  - Facilitate access to affordable diagnosis, treatment and high-quality care, including across borders
  - Promote medical education, research, information dissemination, evaluation of care
- Creation
  - 15/12/2016: *Board of Member States*
  - 01/03/2017: kick off
  - 2022-23: renewal → September 30, 2027
  - Unformal network of Hospitals endorsed by each Member State
- Current landscape
  - 24 European Networks
  - 20 ERNs: rare diseases in the strict sense
  - 3 ERNs: rare cancers
  - 1 ERN for childhood transplantation
  - About 400 hospitals in 25 EU countries + Norway
  - > 1600 centres of expertise, including 229 centres in France



European Reference Networks ITHACA

24 ERNs covering all aspects of rare/complex human diseases + rare cancers

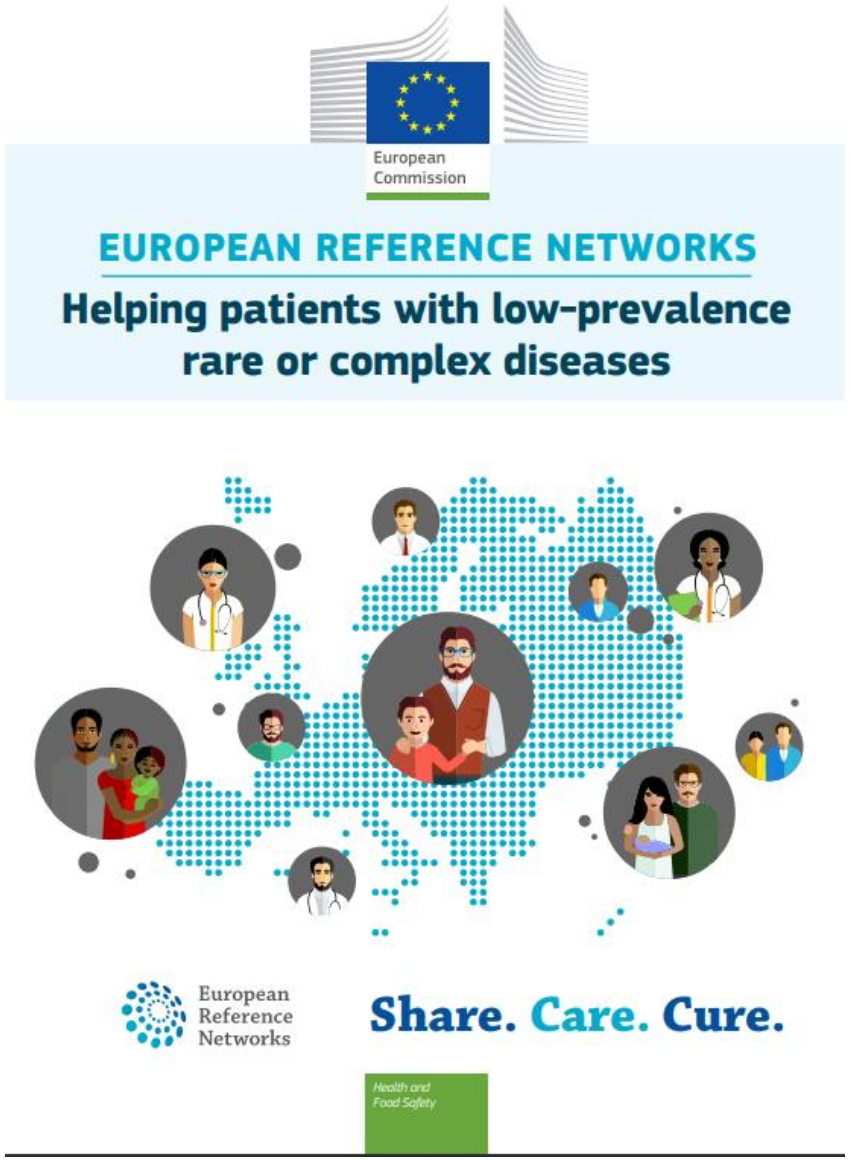
- Organs/systems (12)
  - Gateway to Uncommon And Rare Diseases of the HEART - GUARD-HEART
  - ERN on Respiratory Diseases - ERN-LUNG
  - ERN on Rare Hepatological Diseases - RARE-LIVER
  - European Rare Kidney Diseases RN – ERKNet
  - ERN on Rare Multisystemic Vascular Diseases – VASCERN
  - Rare Endocrine Conditions - Endo-ERN
  - Rare Bone Disorders - ERN BOND
  - ERN on Rare and Undiagnosed Skin Disorders - ERN-Skin
  - ERN on Rare Hematological Diseases - EuroBloodNet
  - ERN on Rare Eye Diseases - ERN-EYE
  - Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases Network - RITA
  - Rare Connective Tissue and Musculoskeletal Diseases Network – ReCONNET
- Developmental anomalies/Genetics (1)
  - **ERN on Developmental and Neurodevelopmental Anomalies - ITHACA**



- Neurology (4)
  - ERN on Rare and Complex Epilepsies - EpiCARE
  - ERN on Rare Neurological Diseases - ERN-RND
  - ERN for Rare Neuromuscular Diseases - EURO-NMD
  - ERN for Rare Hereditary Metabolic Disorders – MetabERN
- Surgery/mixed (4)
  - Rare craniofacial anomalies and ENT disorders - ERN CRANIO
  - ERN on Rare inherited and congenital anomalies - ERNICA
  - Rare Urogenital Diseases – eUROGEN
  - ERN on Transplantation in Children – TransplantChild
- Oncology (3)
  - ERN on GENetic TUmour RIsk Syndromes - ERN GENTURIS
  - ERN on Rare Adult Cancers (solid tumors) - EURACAN
  - ERN for Paediatric Cancer (haemato-oncology) - PaedCan-ERN

European Reference Networks ITHACA

ERNs an inovative BUT complex approch to Health

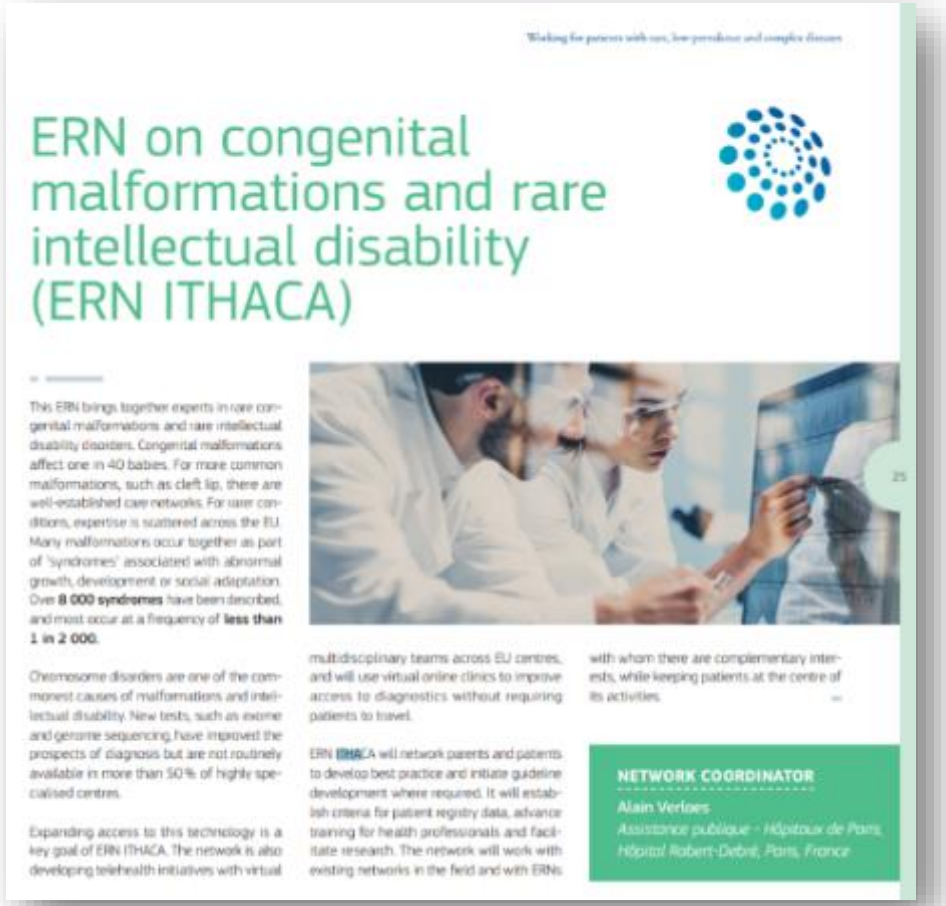


- R = REFERENCE not RESEARCH
- Network of experts in rare/complex disorders
  - Working in national expert centres
  - Coordinated by a coordinator elected by its peers
- ERNs are not legal entities (not EU bodies/infrastructure)
- Based on a network of HCPs harbouring expert centres
  - Proposed by national authorities (no universal criteria)
  - NOT funded by EU for their involvement
  - The coordinating HCP is **funded for coordination** (ca 800 k€/Y) by EC
  - No co-funding requested by HCPs
- Linked to a dedicated Board of Member States
  - Variable interest to RD community
- « Subsidiary principle »
  - At least 27 Healthcare systems (regional OR national approach)
  - 27 research policies & medical cursus
  - Different access and refunding strategies for genetic tests
  - National RGPD constraints

https://ern-ithaca.eu

Intellectual disabilities, TeleHealth, Autism and Congenital Anomalies

- 2017: Coordinated by Manchester (Prof. Jill Clayton-Smith)
- 2019: Coordination transferred to Paris (post-Brexit)
- Network
  - 2017: 38 HCP
  - 2022: 71 HCP



ERN on congenital malformations and rare intellectual disability (ERN ITHACA)

This ERN brings together experts in rare congenital malformations and rare intellectual disability disorders. Congenital malformations affect one in 40 babies. For more common malformations, such as cleft lip, there are well-established care networks. For rarer conditions, expertise is scattered across the EU. Many malformations occur together as part of 'syndromes' associated with abnormal growth, development or social adaptation. Over **8 000 syndromes** have been described, and most occur at a frequency of **less than 1 in 2 000**.

Chromosome disorders are one of the commonest causes of malformations and intellectual disability. New tests, such as exome and genome sequencing, have improved the prospects of diagnosis but are not routinely available in more than 50 % of highly specialised centres.

Expanding access to this technology is a key goal of ERN ITHACA. The network is also developing telehealth initiatives with virtual multidisciplinary teams across EU centres, and will use virtual online clinics to improve access to diagnostics without requiring patients to travel.

ERN ITHACA will network parents and patients to develop best practice and initiate guideline development where required. It will establish criteria for patient registry data, advance training for health professionals and facilitate research. The network will work with existing networks in the field and with ERNs with whom there are complementary interests, while keeping patients at the centre of its activities.

NETWORK COORDINATOR
Alain Verloes
Assistance publique - Hôpitaux de Paris, Hôpital Robert-Debré, Paris, France

ITHACA

European Reference Network
for rare or low prevalence complex diseases
Network Intellectual Disability and Congenital Malformations (ERN ITHACA)

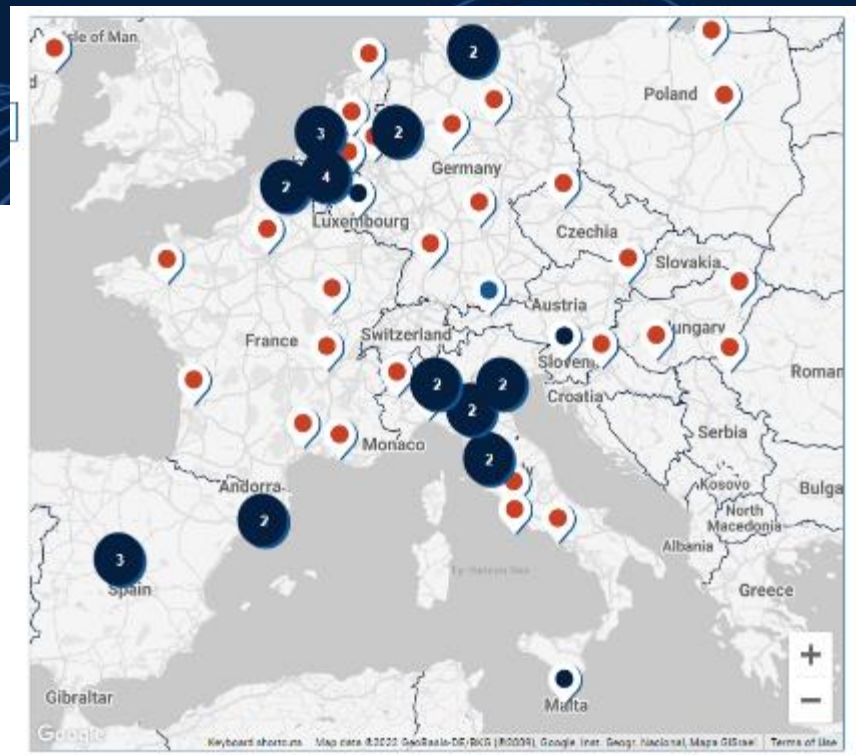
https://ern-ithaca.eu

Intellectual disability, TeleHealth, Autism and Congenital Anomalies

- Landscape
  - Most HCPs are genetics departments in teaching hospitals

- Scope
  - Developmental abnormalities
    - Simple malformations and MCA
    - NDD (ID, ASD)
  - Genetic, multifactorial (e.g. spina bifida) or environmental cause

Intellectual disability, TeleHealth, Autism and Congenital Anomalies



ITHACA

European Reference Network

- History
  - 2017-(2019)-2022 : ERN v1 (cofunding EU-HCP)
  - 2022- 2023: ERN v2 bridge grant (100% EU funding)
  - 1er Octobre 2023 - 30 Septembre 2027: ERN v2 final Grant
  - **3 224 561 € (806 140 /an)**
- Organization
  - Multi-beneficiary
    - F: APHP RDB
    - NL: Amsterdam, Rotterdam, Groningen
    - I: OPBG
  - Coordination team : 10 personnes
    - Paris : 3 PM + Anne (support financier : filière/APHP)
    - Groningen, Amsterdam, Rotterdam, Berne, Rome
  - ExCom
    - coordination + WP leads
- Board
  - 71 HCP de 25+1 countries, including 3 hubs
  - Related Structures: ERDERA (EJPRD+ Solve RD), Orphanet, IRdIRC, JARDIN
- Non-EU affiliated partners (in progress)
  - National Consortia : Switzerland, Turkey, Ukraine
  - UK (Manchester, Others ?)
  - **Extra European ??**

Governance

ERN Board

Member state 1
- HCP1-1 Medical Team — Coord
- HCP1-2 Medical Team — Coord

Member state 2
- HCP2-1 Medical Team — Coord
- HCP2-2 Medical Team — Coord

Member state n
- HCPn-1 Medical Team — Coord
- HCPn-2 Medical Team — Coord

Executive Committee

- EJP-RD — 1 invited rep
- ERDERA — 1 invited rep
- Orphanet — 1 invited rep
- JARDIN
- IRDiRC — 1 invited rep

Coordination team

ERN Coordinator

PM supporting WGs

- ERN PM
- PM Patient-oriented Actions & CPMS
- Communication Manager and Webmaster
- Pedagogical engineer - APOGeE
- Data Manager – ILIAD
- Methodoligist CPGs and CDSTs (NL)
- Project Officer – CPGs and CDSTs (NL)
- Project Officer – NDDs (NL)
- Project Officer – Research (IT)
- IT developer (NL)
- DataManager – SysNDD (CH + DE)

Work Packages

- WP1 Coordination
- WP2 Dissemination
- WP3 Evaluation
- WP4 CPMS
- WP5 Registries
- WP6 T&E
- *WP7 Guidelines*
- *WP10 Research & Pt*
- WP8 Ukraine (1)
- WP9 Ukraine CPMS

Working Groups

- **WG1** Coordination
- **WG2** Dissemination
- **WG3** Evaluation
- **WG4** CPMS Healthcare — Task 1, Task 2, Task 3, Task n
- **WG7** ILIAD registry — Task 1, Task 2, Task 3, Task n
- **WG8** Teaching & Training — Task 1, Task 2, Task 3, Task n
- **WG9** APOGeE — Task 1, Task 2, Task 3, Task n
- **WG11** Expert Cons Statements — Task 1, Task 2, Task 3, Task n
- **WG12** Research & Innovation — Task 1, Task 2, Task 3, Task n
- **WG5** NeuroDev Disorders — Task 1, Task 2, Task 3, Task n
- **WG14** Patient Adv Brd
- **WG15** ELSI Adv Board
- **WG10** EU Certif — Task 1, Task 2, Task 3, Task n
- **WG6** Fetal medicine — Task 1, Task 2, Task 3, Task n
- **WG13** SB -Fetal surgery — Task 1, Task 2, Task 3, Task n

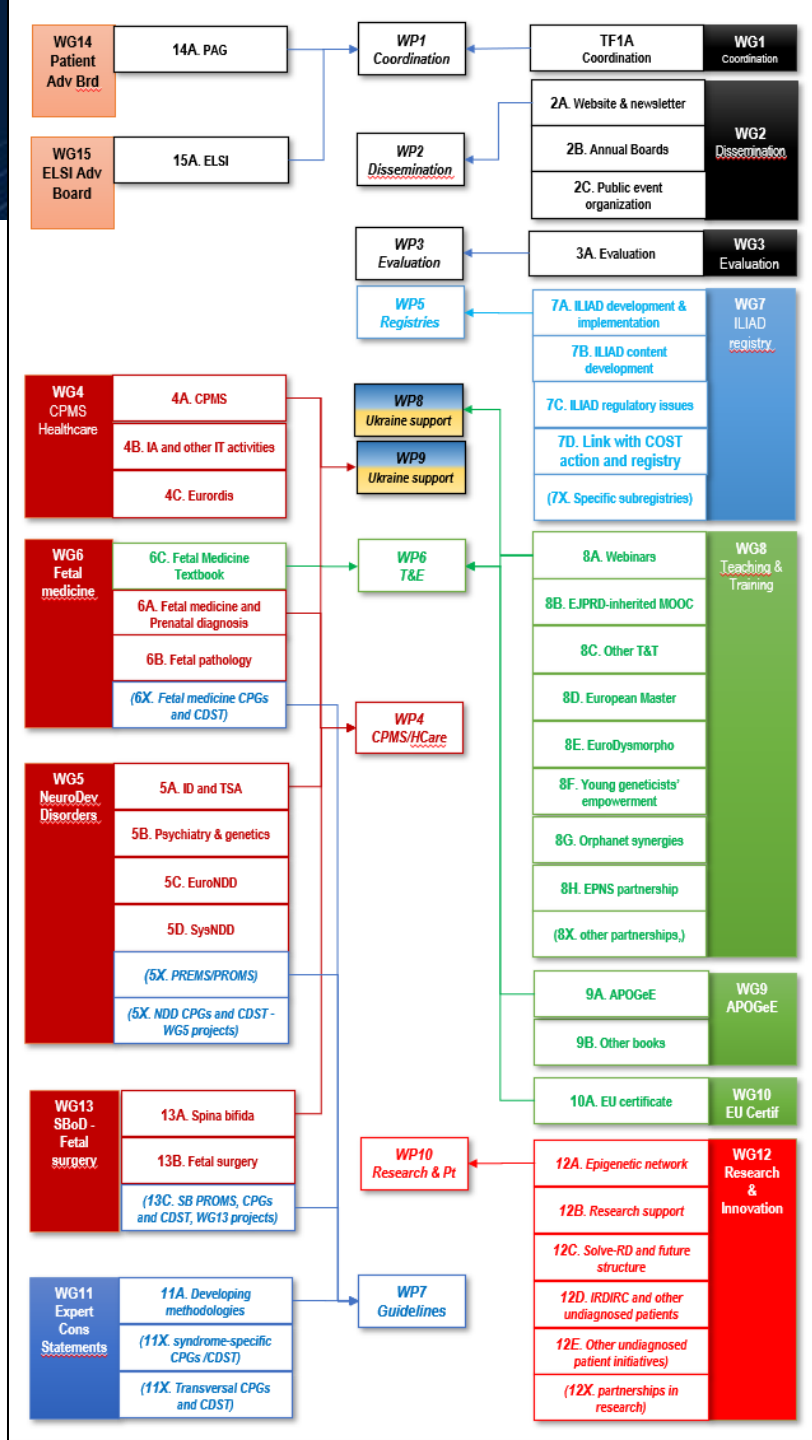
WG chair + 1 co-chair → ExCom

- **WG chair** — Co-chair 1, Co-chair 2, Co-chair 3, Co-chair n
- **Governance Of WGs**

WP, WG et Task Forces

- 15 Working Groups consisting in :
- 39 permanent Task Forces
- + ~10 TF ad hoc
  - for specific thematic tasks (Guidelines, ITHACA sub-registers, PROMS))

- Connexions
  - ERDERA (European Rare Diseases Research Alliance) :
    - EU ResearchCoordination / MR
    - JARDIN: implementation of national RD networks in Europe
  - ERICA: Common regulatory documents

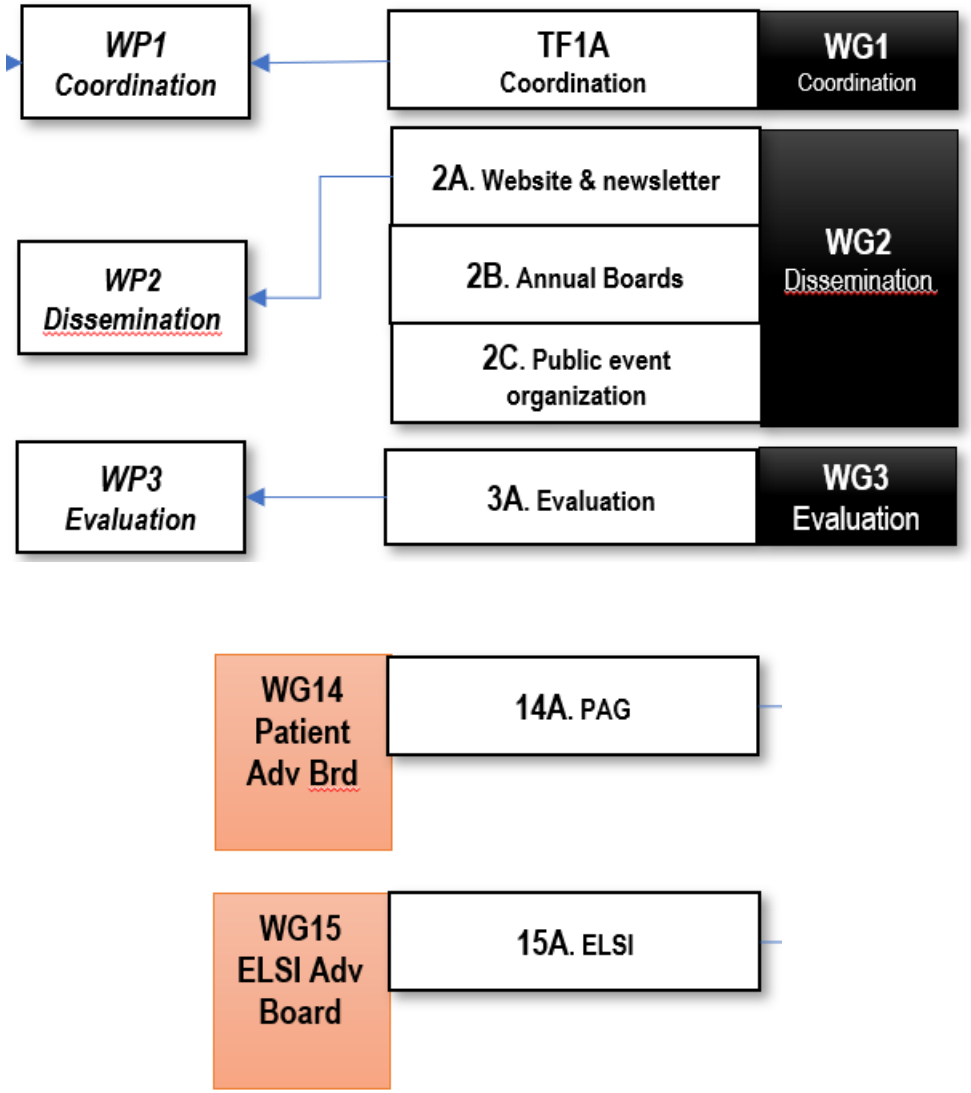


New axes for 2023

- Fetal Surgery for Dysraphism
- Psychiatric manifestations of monogenic diseases
- Undiagnosed patients
- PROMS

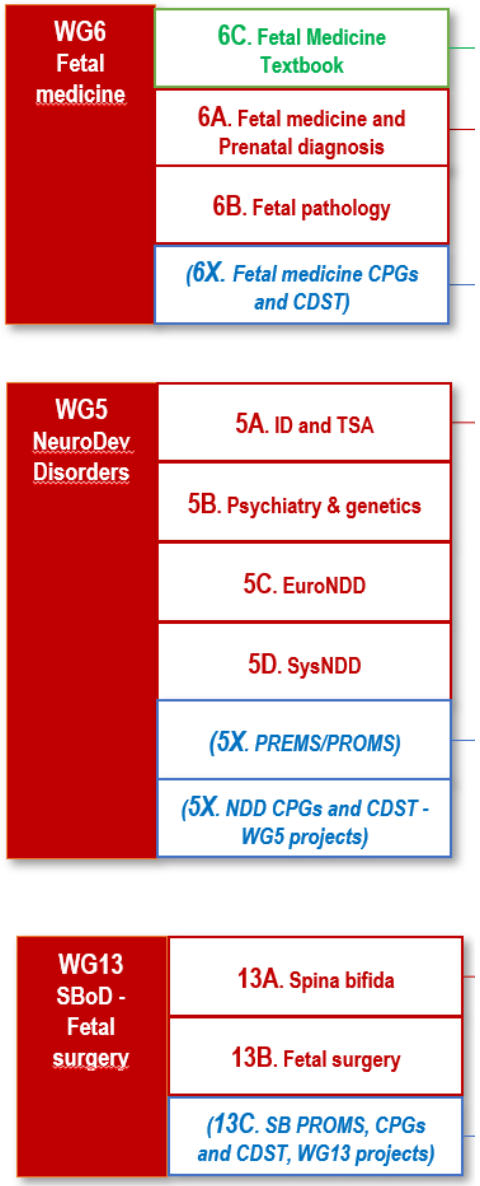
European Reference Networks ITHACA

WP : general governance and advisory bodies



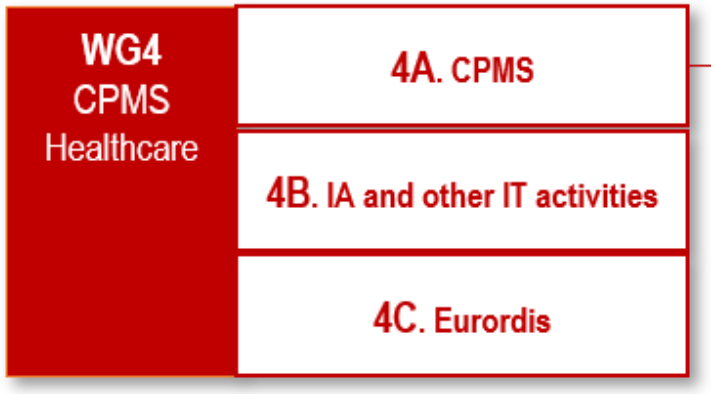
- General coordination
  - Boards & Excoms
  - Support to events
  - Website & NL
  - Reporting
- Relations with stakeholders & funding agencies
  - DGSante (EC governance)
  - HADEA (finance, EU level)
  - APHP (finance, day-by-day)
  - ERDERA
- Patients advocacy groups
- External ethical committee

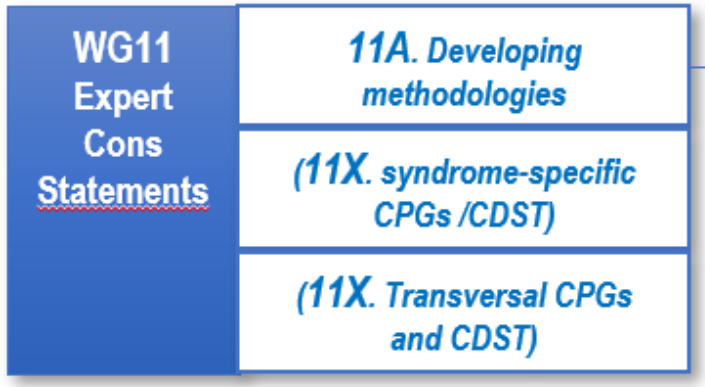
WP: clinical orientation



- Neurodevelopmental disorders
  - Workshop
    - EuroNDD
  - Web resource
    - SysNDD
  - Guidelines (trans-WG)
  - PROMS
  - Psychiatric aspects
- Developmental disorders
  - Fetal medicine and PDg
  - Fetal pathology
    - Winter course
  - Dysraphisms (trans-ERN)
  - Fetal surgery

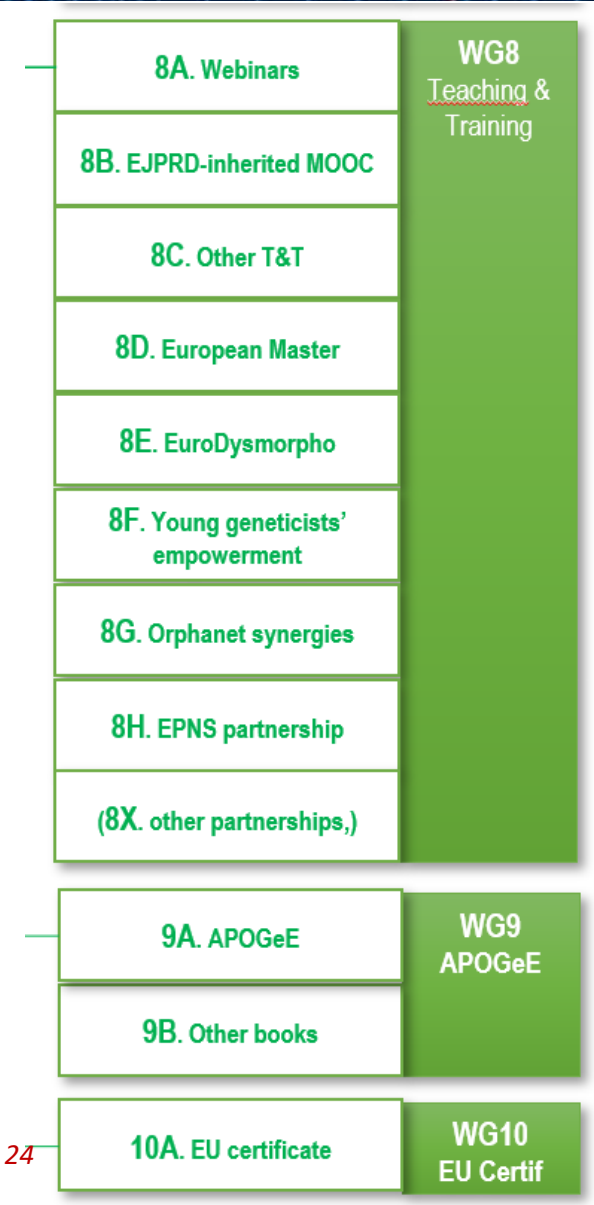
WP: experts & expertise





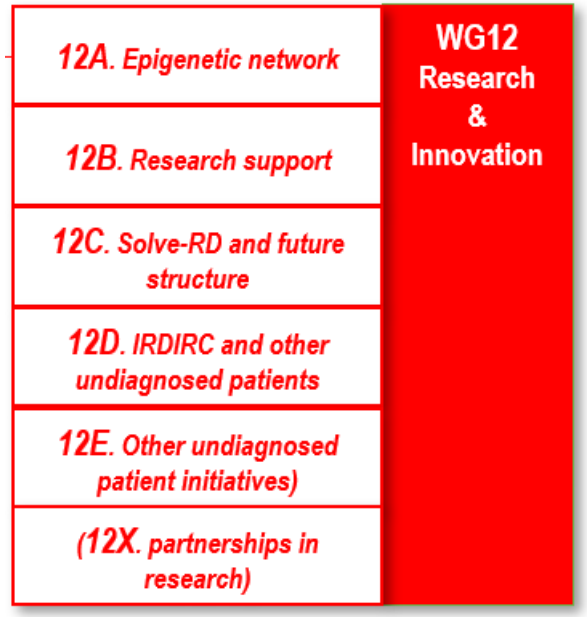
- CPMS
- Expert statements
  - Clinical Practice Guidelines (CPGs)
    - Syndrome specific
    - Problem specific
    - Transversal
  - Clinical Decision Support Tools (CDST)
  - Endorsement of existing CDST/CPG
  - Methodology : AGREE II + Delphi
- PROMS
  - With ERDERA

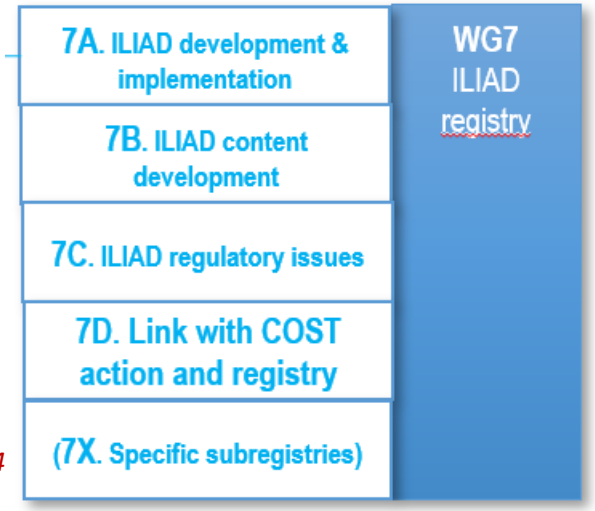
WG : e-Education



- Webinars
- MOOCS
- APOgeE and other online teaching/learning material
- Support to EU UEMS certificate of genetics

WG: research support



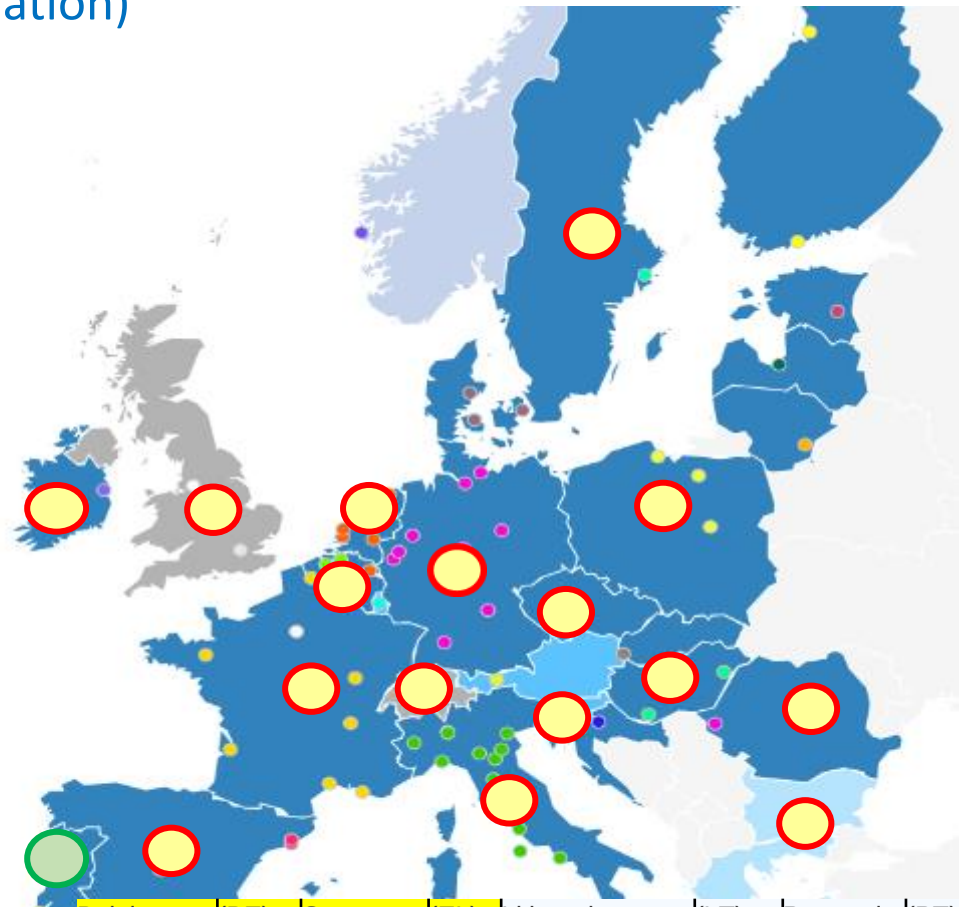


- Clinical research calls (series of patients)
  - Trough web site
  - Free access to the scientific community
- Short exchanges (with EJPRD)
- Contribution to Solve-RD and ERDERA
- Support to IRDiRC
- ILIAD registry
  - Core database on monogenic ID patients
  - Disorder-specific databases

Une communauté de patients en pleine croissance - ePAGS (38 ePAGS + 30 PC)

- Ouverture aux Assos sans diagnostic (SWAN, UDNI, Wilhelm Foundation)

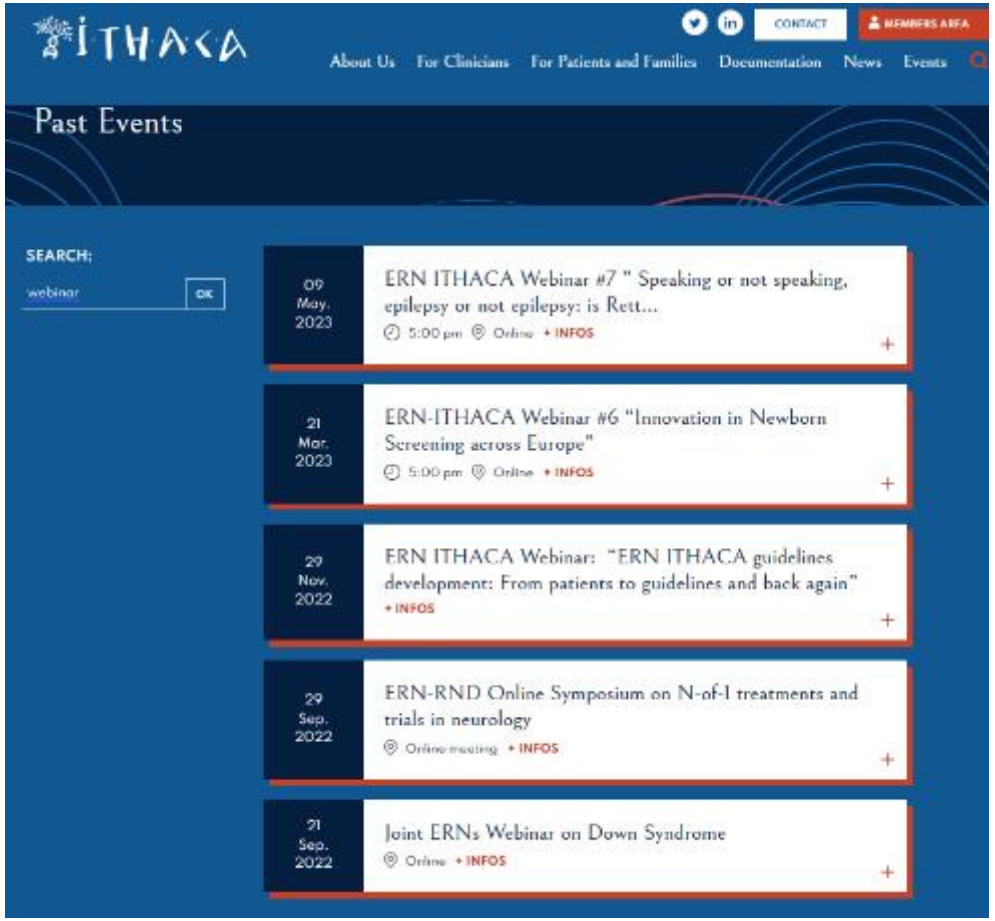
22q11.2	Acrodysostosis	ADNP	Angelman	Beckwith-Wiedemann	Chromosomal disorders
Chromosomal abnormalities	Corpus Callosum	Costello and cardio-facio-cutaneous syndrome	Cowden (mental health)	Coffin-Lowry	Cdls Cornelia Delange
CDKL5	Dravet	Fragile X	Goldenhar	Kleefstra	MED13L
Mowat-Wilson	Noonan	PACS1	Pallister-Killian	Pitt Hopkins	Prader Willi
Phelan Mc Dermid	Rett	SAT2B	Smith-Lemli-Opitz	Smith-Magenis	Spina Bifida
Tuberous Sclerosis Complex	Williams	White-Sutton	Brain malformations, genetic on NDD	ID rare diseases	Ultra rare condition
	Undiagnoses no names	RD Borys the Hero Foundation	RD Alliance IT,ES,CZ,NL,FR,RO	Etc ..... Specific conditions	



Belgium	(BE)	Greece	(EL)	Lithuania	(LT)	Portugal	(PT)
Bulgaria	(BG)	Spain	(ES)	Luxembourg	(LU)	Romania	(RO)
Czechia	(CZ)	France	(FR)	Hungary	(HU)	Slovenia	(SI)
Denmark	(DK)	Croatia	(HR)	Malta	(MT)	Slovakia	(SK)
Germany	(DE)	Italy	(IT)	Netherlands	(NL)	Finland	(FI)
Estonia	(EE)	Cyprus	(CY)	Austria	(AT)	Sweden	(SE)
Ireland	(IE)	Latvia	(LV)	Poland	(PL)		

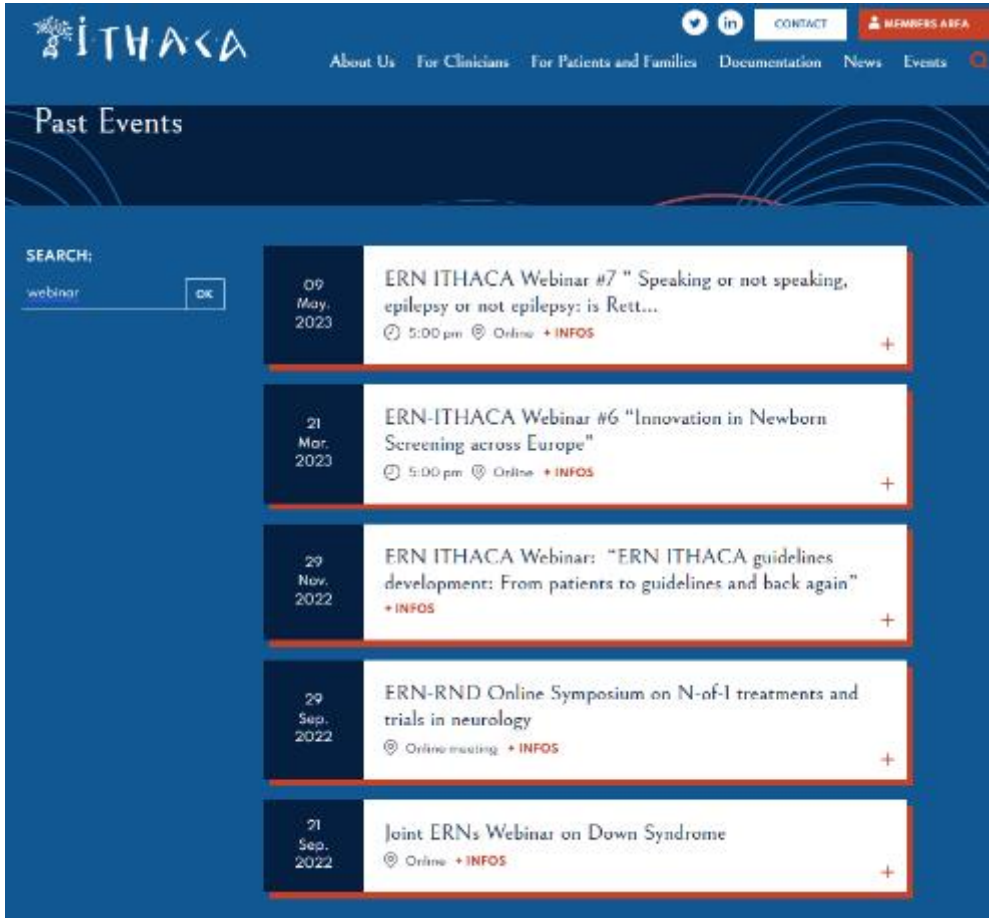
United Kingdom

WG8: Teaching & Education - Webinars



- Coordination: Laurence FAIVRE (Dijon, FR) & Guiseppe ZAMPINO (Rome, IT)
- Easy support for e-learning
  - ZOOM, TEAMS…
  - Synchronous & interactive… OR not
  - Advertisement through ERN NL and Organizing partners
  - Usually opened to participants outside the Network
  - Further dissemination through dedicated ERN YouTube channel
- From some webinars to > 10/ERN/year
- Common webinars with EPNS !!!

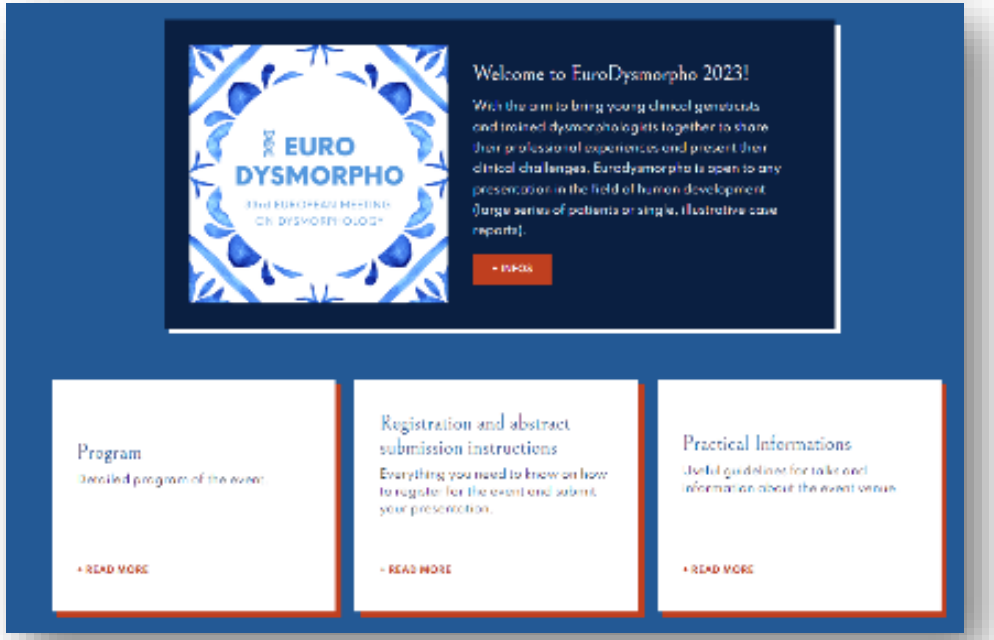
WG8: Teaching & Education - Webinars



- Coordination: Laurence FAIVRE (Dijon, FR) & Guiseppe ZAMPINO (Rome, IT)
- Easy support for e-learning
  - ZOOM, TEAMS…
  - Synchronous & interactive… OR not
  - Advertisement through ERN NL and Organizing partners
  - Usually opened to participants outside the Network
  - Further dissemination through dedicated ERN YouTube channel
- From some webinars to > 10/ERN/year
- Common webinars with EPNS !!!

WG8: Teaching & Education - Eurodysmorpho

- A 3-day workshop dedicated to developmental anomalies
  - > 80 oral presentation
  - 5 educational sessions
- Oriented to (and organized with) Young Geneticists and Young Child Neurologists



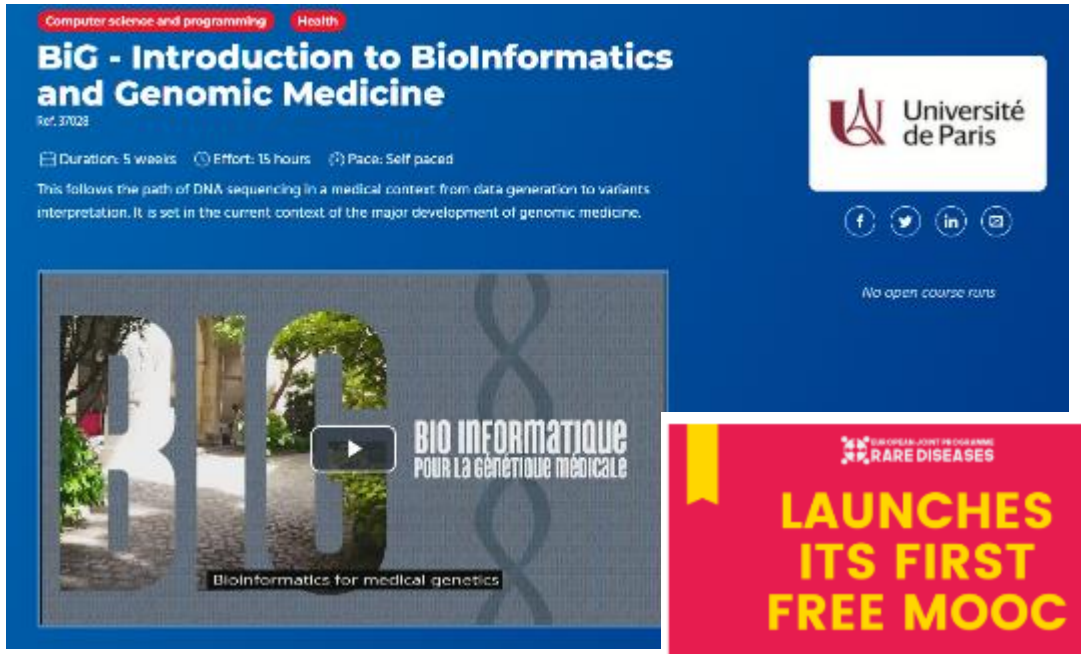
- 2024: Ljubliana
- 2025: Vilnius
- 2026: Italia

European Reference Networks ITHACA

WG8: Teaching & Education - MOOCS and other self-learning tools

- Asynchronous or synchronous (MOOCS) e-learning tools
  - BIG
    - Developped with Université Paris Cité
    - Second run in December 2023
  - Diagnosing Rare Diseases: from the Clinic to Research and back

    Second run from October 4th
    - EJPRD→ ITHACA in Dec 2023
    - 5 224 enrolled on this course
    - 5 weeks - 3 hours per week
    - Digital certificate when eligible

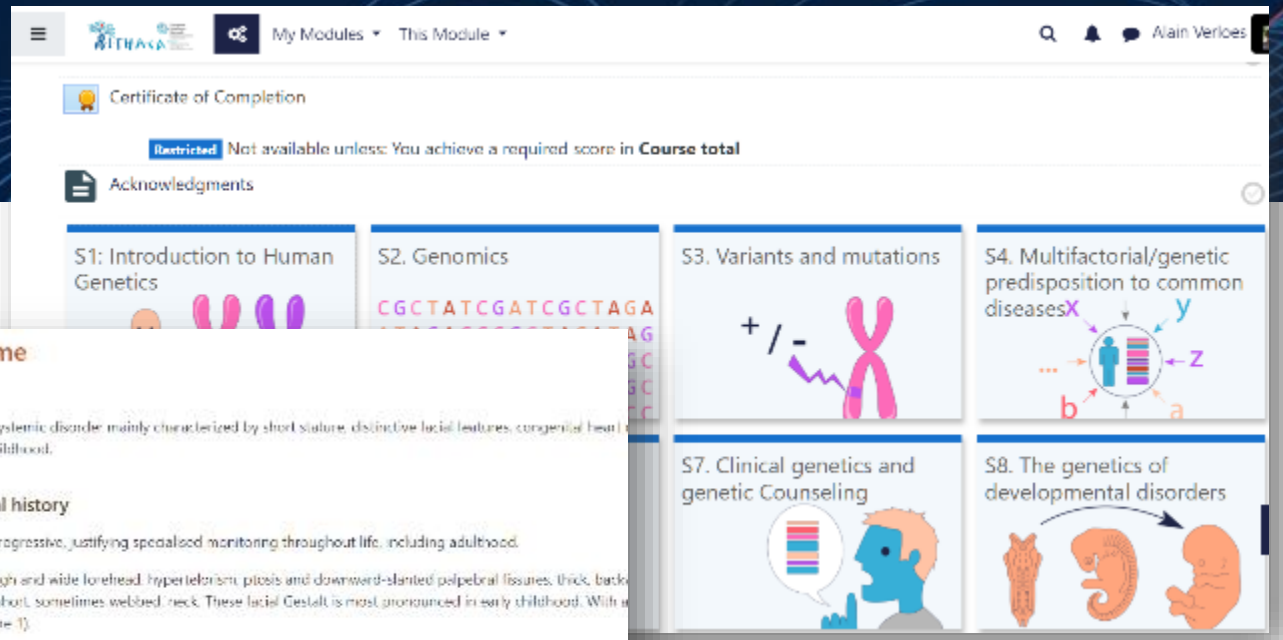


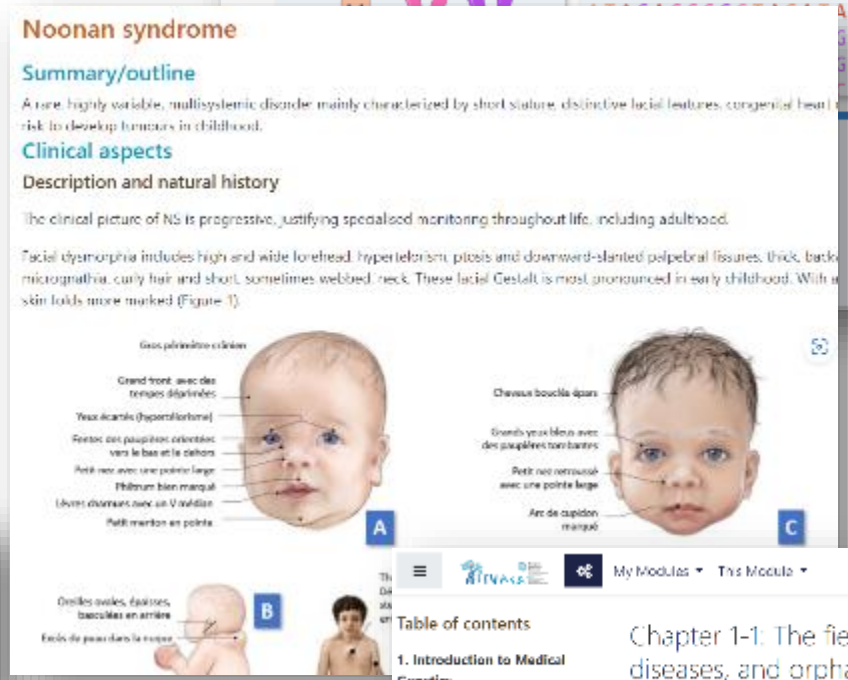
- From focused information

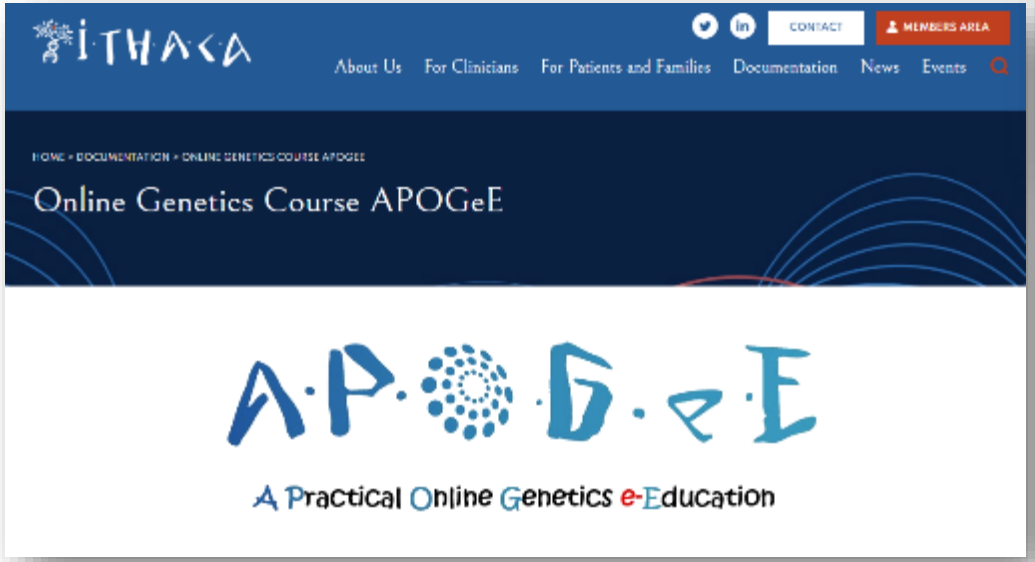
  …to large e-textbooks :
  - ITHACA: APOGeE

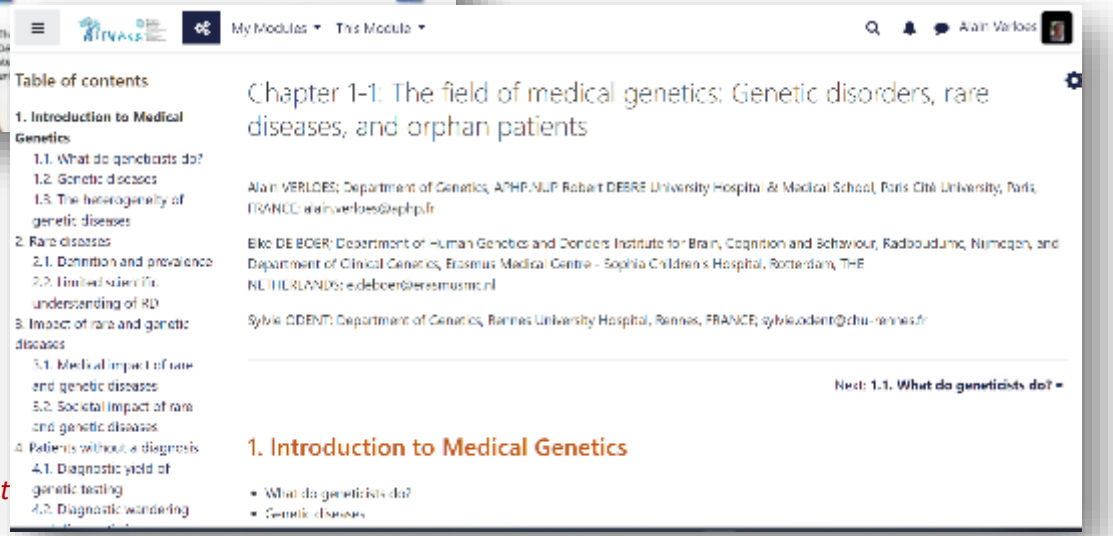
WG9: APOGeE

- Coordination: A Verloes + steering committee
- APOGeE
  - Online e-handbook on medical genetics
  - 100 contributors
  - Adopted as reference for **ECCMG certificate for Med Genetics** (UEMS)
  - With strong accent on NDD and developmental anomalies
  - In development (beta version)
  - Achievement T2, 2024







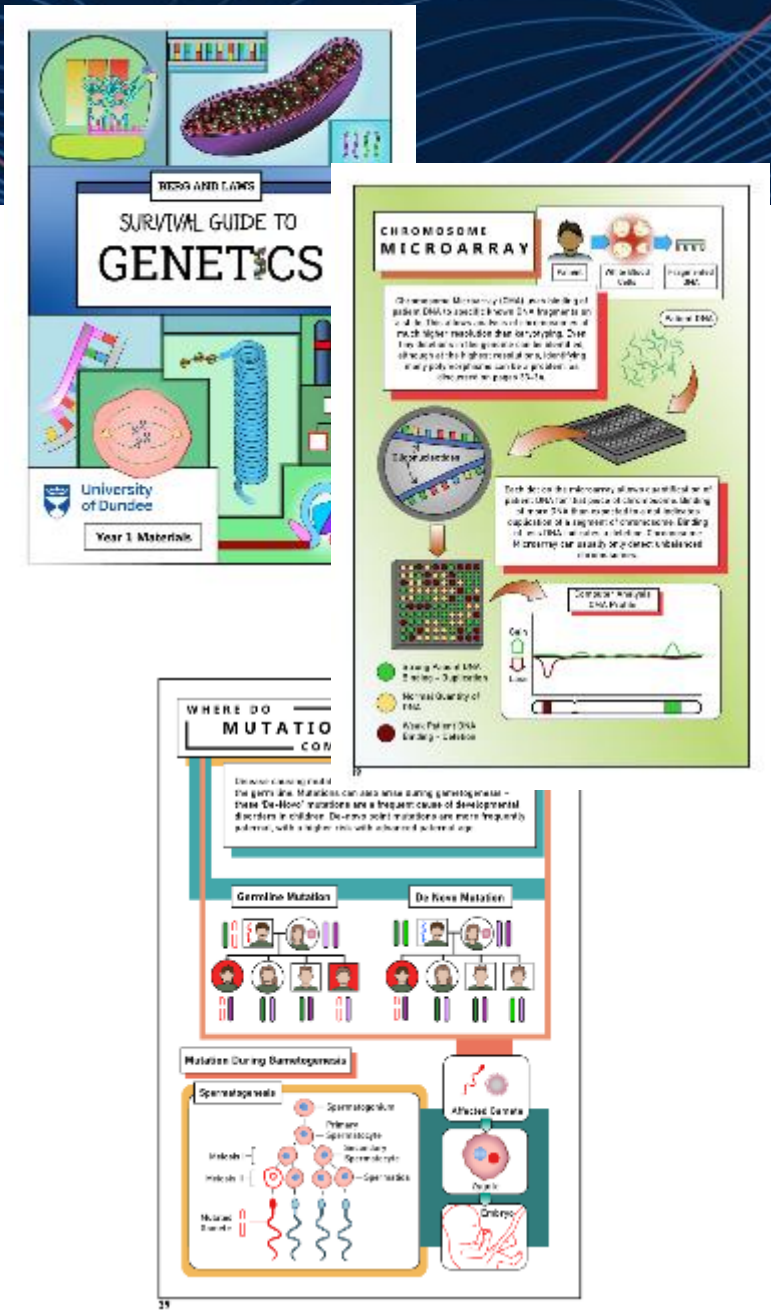


WG9 : other supports

- Dysmorphology Vade Mecum
  - Adaptation in GB of a French book (foreseen : mid 2024)

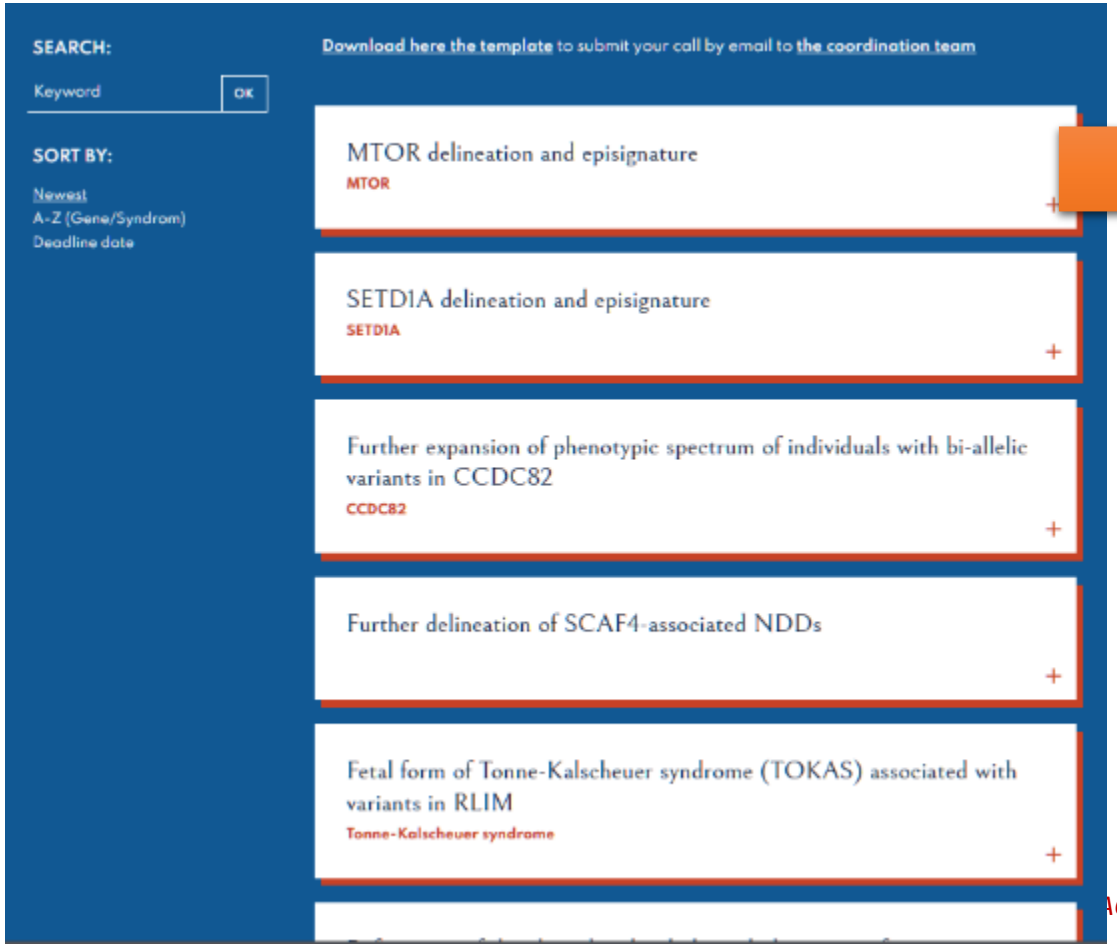
- Genetic Survival Guide
  - Small graphic book < Dundee University: VO online July 23
  - To be adapted in several languages(FR to begin)
  - Planned: "Genetic counseling" in the same format

- Other projects
  - Fœtal pathology textbook?



WG13: research support - Research calls

- Open to ITHACA and beyond
- Clinical research calls (series of patients)



Targeted gene under study

Smith-Kingsmore syndrome MTOR germinal (OMIM 616638 , ORPHA 457485)

Abstract

Together with the Dijon team, we aim to better define the clinical, psychiatric and neuropsychological characteristics of people with germline MTOR variants, and to describe an epigenetic signature. We would be delighted to collect clinical data, neuropsychological, brain MRI, growth curves and a DNA sample. The Montpellier IRB is in the process of notifying us.

Coordinating clinicians/researchers

David Genevieve – d-genevieve@chu-montpellier.fr

Institution

Centre Hospitalier Universitaire de Montpellier, France

Specific requirements beyond clinical data and genotype data sharing:

- Re-analysis of DNA samples: **No**
- Resampling of patients: **No**
- Linked to a translational/basic research project: **No**

- Thank you !

Contacts: alain.verloes@aphp.fr // info@ern-ithaca.eu

RD in France : the 3 RD plans

PNMR 1

2004 – 2008

Create Centres (CR & CC)
RDs became a public health issue
131 labelled RD Centres
National RD Database

PNMR 2

2011 – 2014 (16)

Structure of RD Networks
Networks (*filières*)
Rare Disease Foundation

PNMR 3

2018 – 2022

Consolidate
National RD database mandatory
Undiagnosed patients

PNMR 4

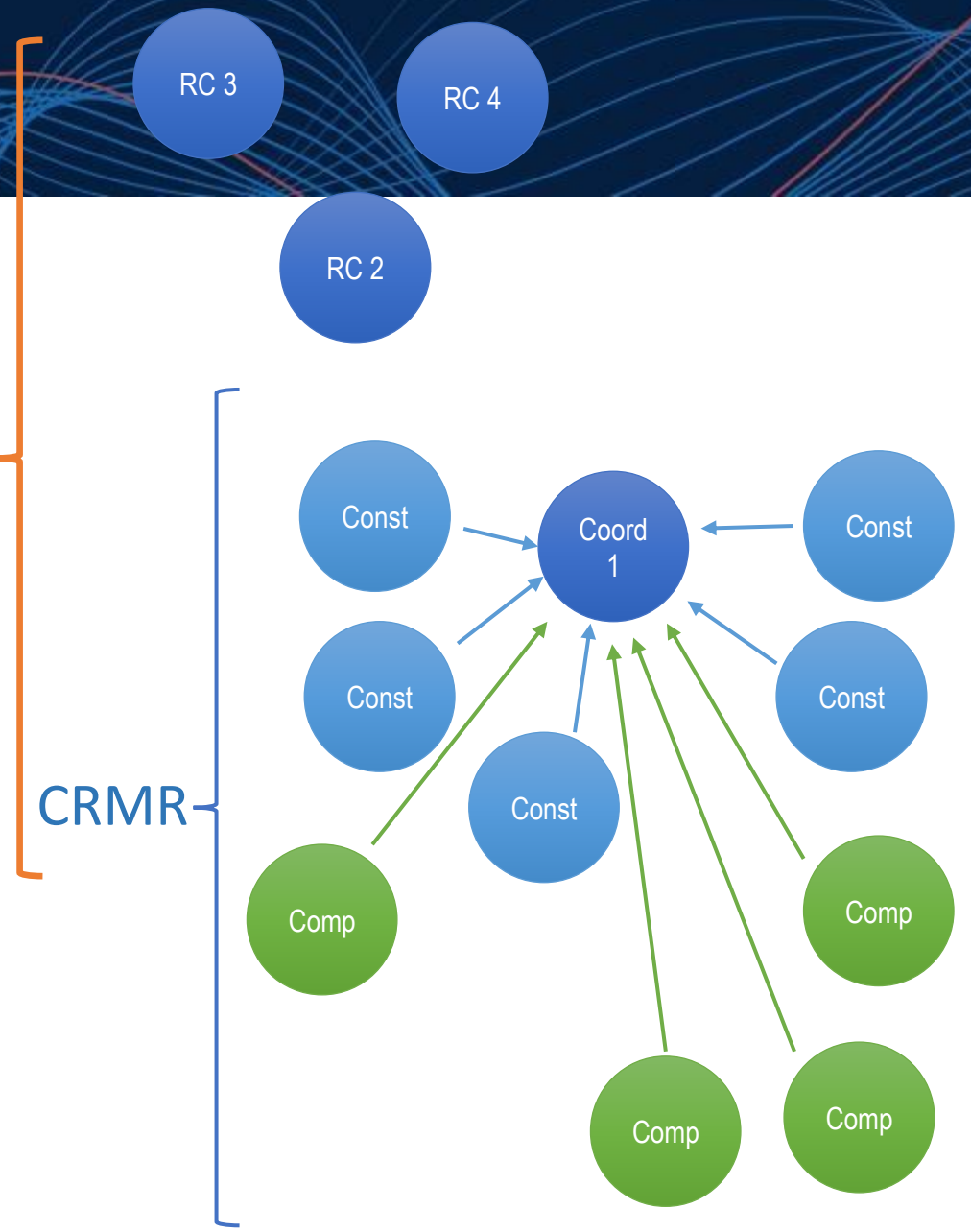
2024 – 2028

Therapies
Health databases
Bioinformatics
?

Concept of RD Reference Centres in France

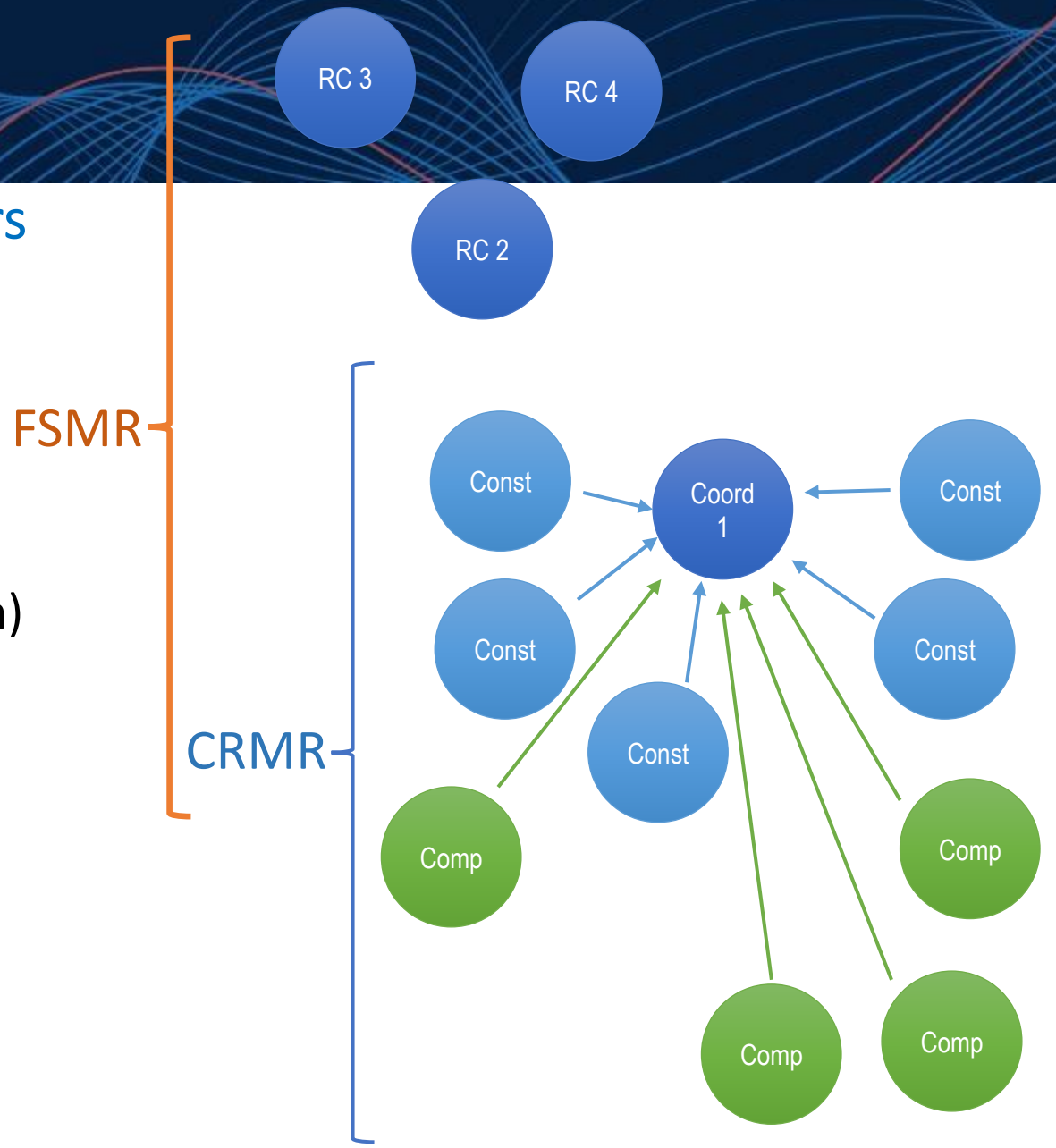
- Holistic organisation for care and cure of rare diseases centered on Hospitals, encompassing local care and family support groups

- Basis: 3 levels
  - 1. Health network of Ref Ct (*Filière de Santé : FSMR*)
    - By broad medical area
    - 1 RC is FSMR coordinator
    - Encompass several Reference Centres
  - 2. Reference Centre (*CRMR*)
    - Monocentric : 1 coordinator
    - Multicentric : 1 coordinator + n constitutive
    - 1 or several in the same theme (depending on the prevalence)
  - 3. Competence centres (*CCMR*)
    - Affiliated with 1 RC
    - Regional mirrors > nation-wide coverage



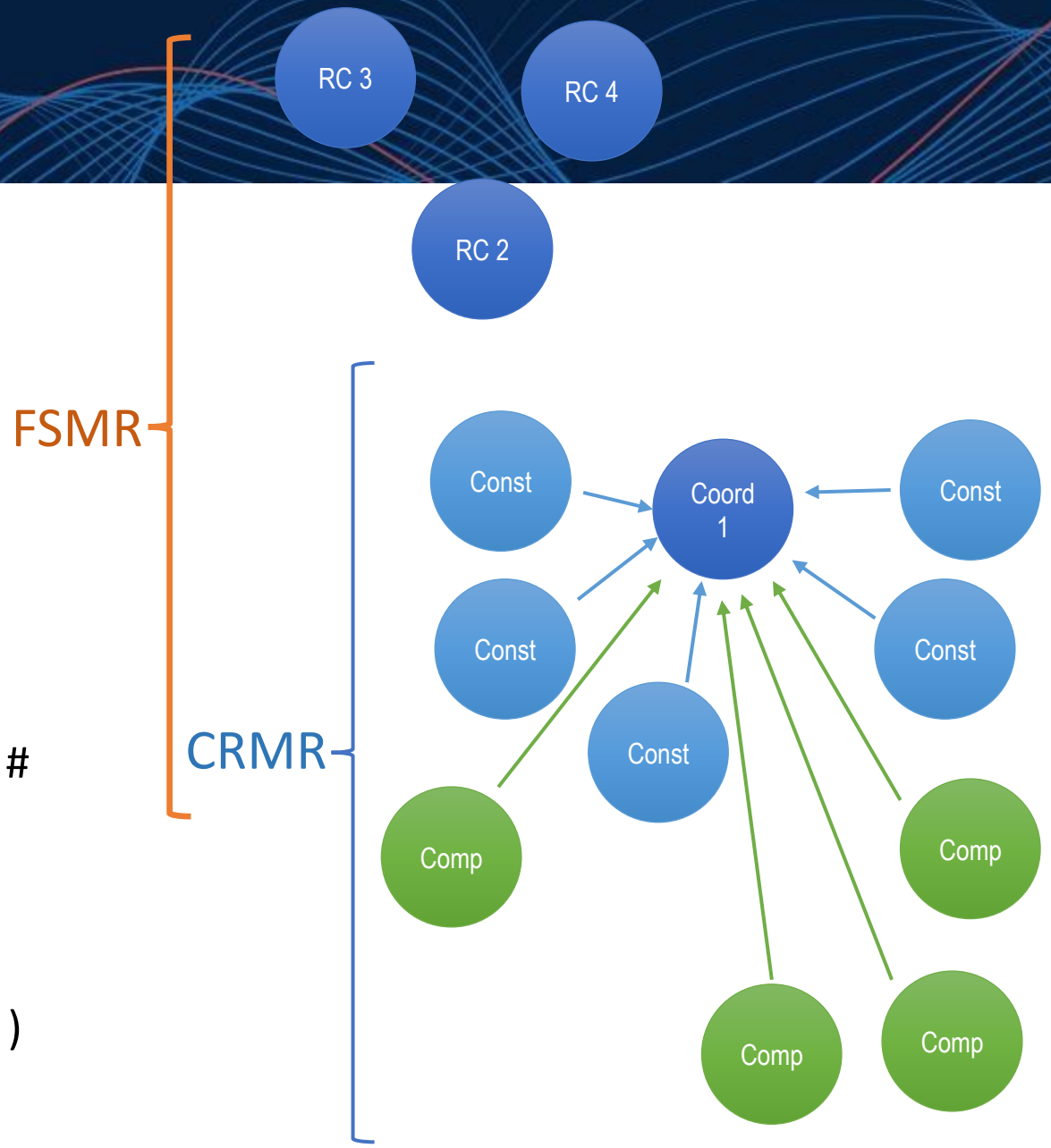
Basic criteria

- Endorsement by Ministry of Health for 5 years
  - Based on a medical project : structure and objectives
  - Evaluation by an independent Jury
- Reference centre
  - # of patients/Y (> 150) – declared through a national Registry
  - 1 or more by field (based on targetted population)
  - Active scientific activity
    - Publication (justified by bibliometry report)
    - PI in at least 1 funded research program (coordonnator or local PI)
  - Involvement in academic teaching
  - Supportive PAGs
- Competence centre
  - # patients/Y (> 25) – declared through registry
  - Geographic coverage complimentary to Ref
  - Active scientific activity
  - Supportive PAGs



Finance

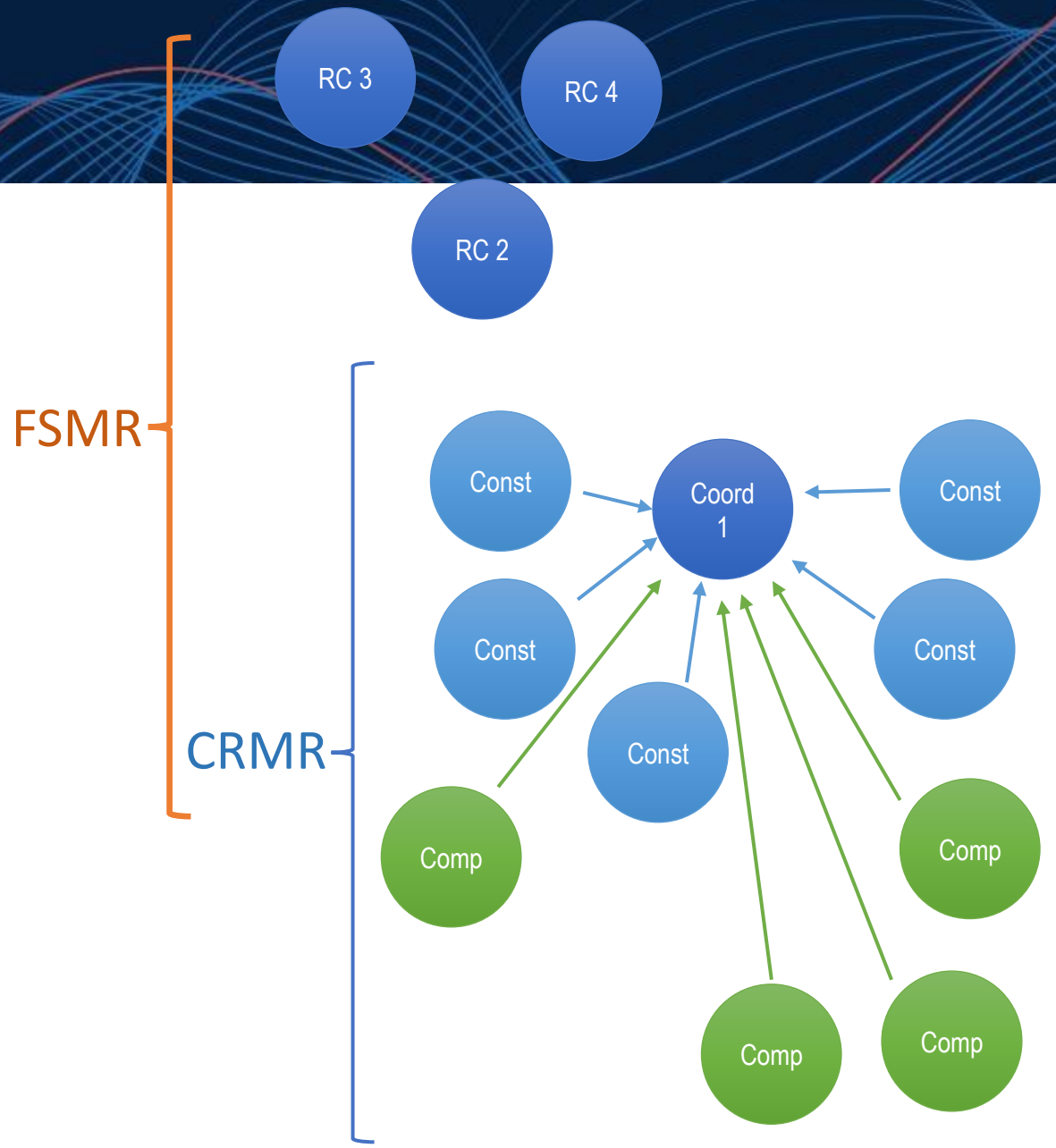
- Closed envelope scattered among partners
  - 260 M/Y
- Health network
  - Coordination of several CRMR
  - FINANCING: for networking activities
- Reference centres
  - A fixed part / hospital + variable part based mainly on # of patients (250-400 k€)
  - Extra financing for the coordinating hospital (60 k€)
  - For non-medical staff and other actions (cannot be used to appoint MD)
  - Hospital overhead = 19% (or more: no commitment !!)
- Competence centre
  - NO FINANCING



Monitoring

- Annual budget established for 5 years

- Annual monitoring
  - FSMR level
    - Activity report (annual)
  - CRMR level
    - Yearly on-line activity report
      - Active file as declared in BaMaRa (justified)
      - + scientific production, teaching, guideline…
      - Hospital impact



RD reference Centres: 5 aims

- Coordination
  - Identifies, **coordinates** and animates its partners (medical, medicosocial, medico-educative)
  - **Integrates PAGs** into the centre's activities and involves them in the definition of its objectives
  - All CRMR actions in coordination with the general objectives of its FSMR
- Expertise
  - Multidisciplinary synthesis or consultation meetings (consensus meetings)
  - Produces National Diagnostic and Care Protocols (*PNDS*)
  - Regular epidemiological records (All patients recorded in the National Rare Diseases DataBank *BNDMR*)
- Remediation
  - Ensures multidisciplinarity, diagnostic, therapeutic and follow-up care
  - Depending on the case, CRMR provides this comprehensive care or organizes it within caregivers
- Research
  - Promotes, animates or participates in translational, clinical, or organizational research.
- Education and training
  - University, postgraduate and other teaching in the field of the group of RDs
  - Therapeutic Education

Filières

23 Filières

- Anomalies du développement déficience intellectuelle de causes rares
- Maladies rares à expression motrice ou cognitive du système nerveux central
- Maladies cardiaques héréditaires
- Maladies rares du développement cérébral et déficience intellectuelle
- Maladies auto-immunes et auto-inflammatoires systémiques rares
- Maladies vasculaires rares avec atteinte multi-systémique
- Maladies rares neuromusculaires
- Maladies rénales rares
- Maladies constitutionnelles rares du globule rouge et de l'érythropoïèse
- Maladies hémorragiques constitutionnelles
- Mucoviscidose et affections liées à une anomalie de CFTR
- Complications neurologiques et sphinctériennes des malformations pelviennes et médullaires rares
- Os-Calcium/Cartilage-Rein
- Maladies hépatiques rares de l'enfant et de l'adulte
- Sclérose latérale amyotrophique et maladies du neurone moteur
- Maladies rares en dermatologie
- Maladies rares endocriniennes
- Malformations abdomino-thoraciques
- Maladies héréditaires du métabolisme
- Maladies rares immuno-hématologiques
- Maladies respiratoires rares
- Maladies rares sensorielles
- Maladies rares de la tête, du cou et des dents

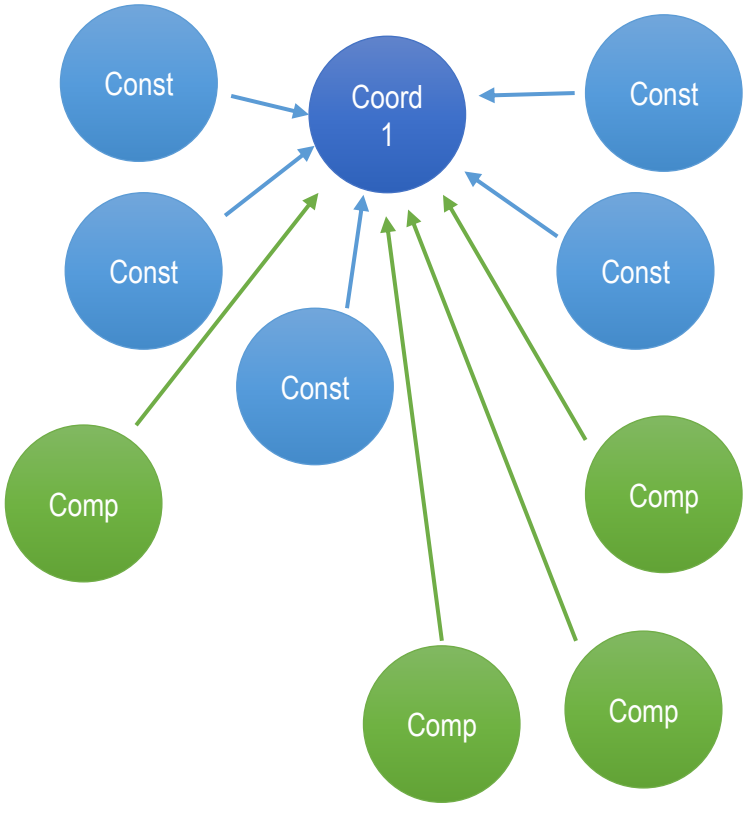
FSMR

RC 3

RC 4

RC 2

CRMR



European Reference Networks ITHACA

Filières

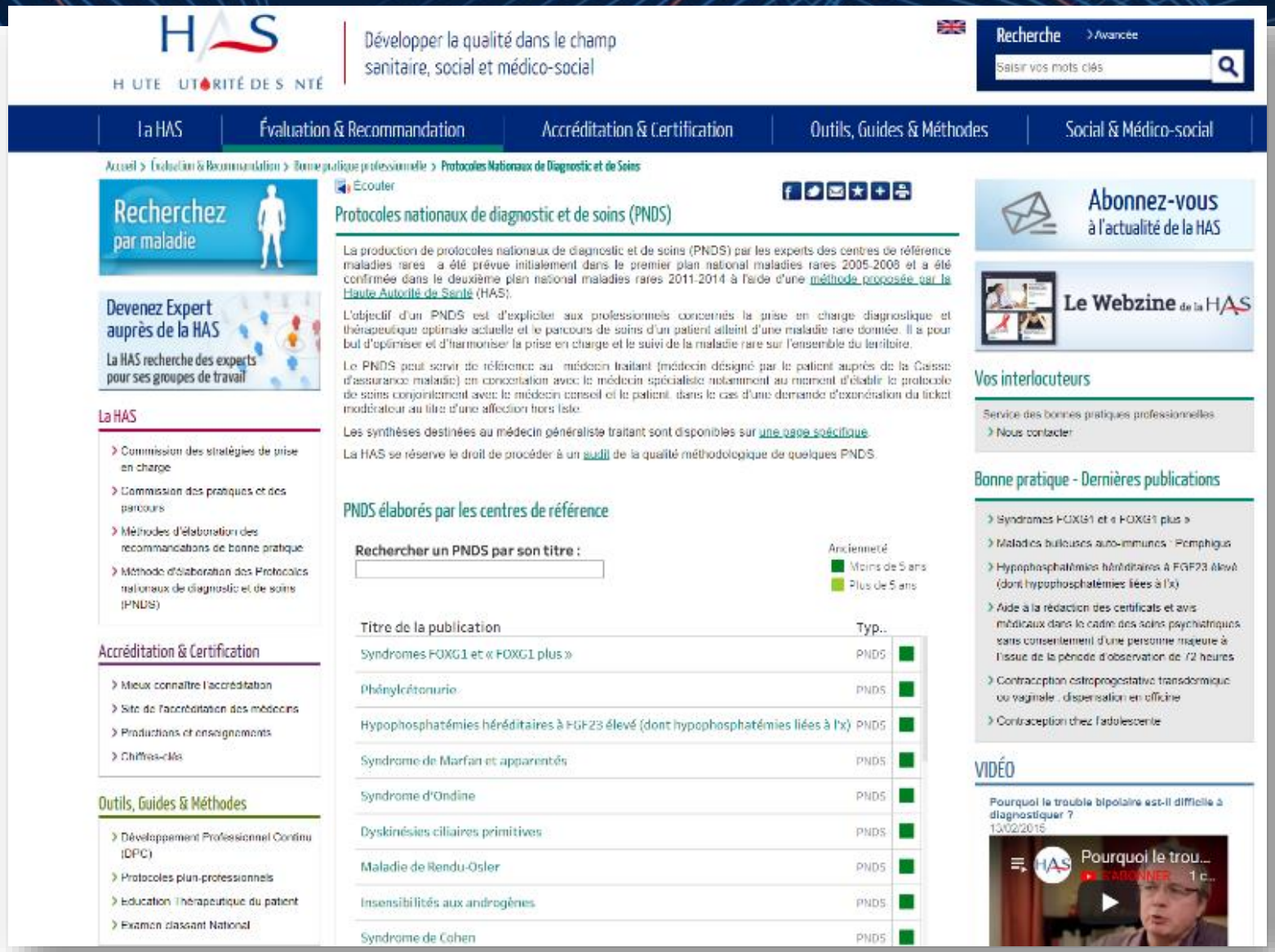
Aims

- promote the legibility of the organization of the RD care to facilitate the orientation of patients and professionalsls
- decompartmentalize (social, medico-social, social)
- Missions

- Improving the care of people with RD
  - The FSMR reinforce, pool and coordinate all actions to improve overall management and the dissemination of expertise and knowledge.
  - The FSMRs do not organise the individual care of patients, which remains the responsibility of the CRMRs attached to them.
- Implementation of translational, clinical and organizational research
  - Support function
- Development of training and information
  - FSMRs identify and support educational programs
  - Information for FSMR stakeholders and outside the FSMR

Centre mapping

National Protocol for Diagnosis and Care

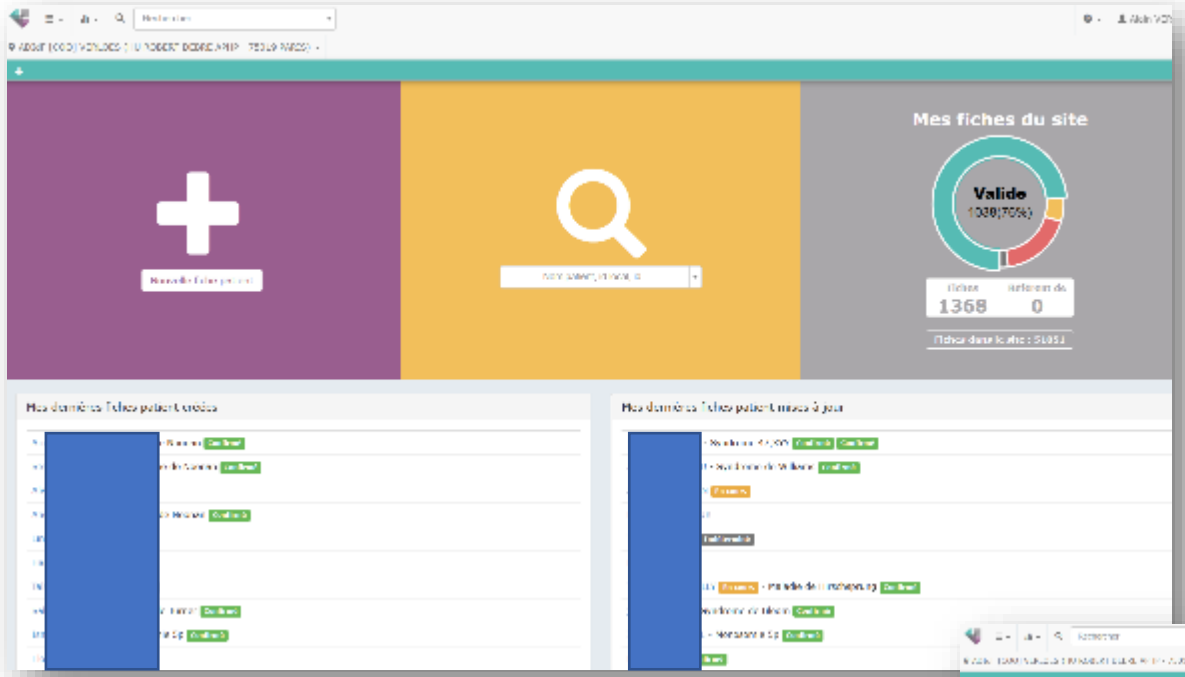


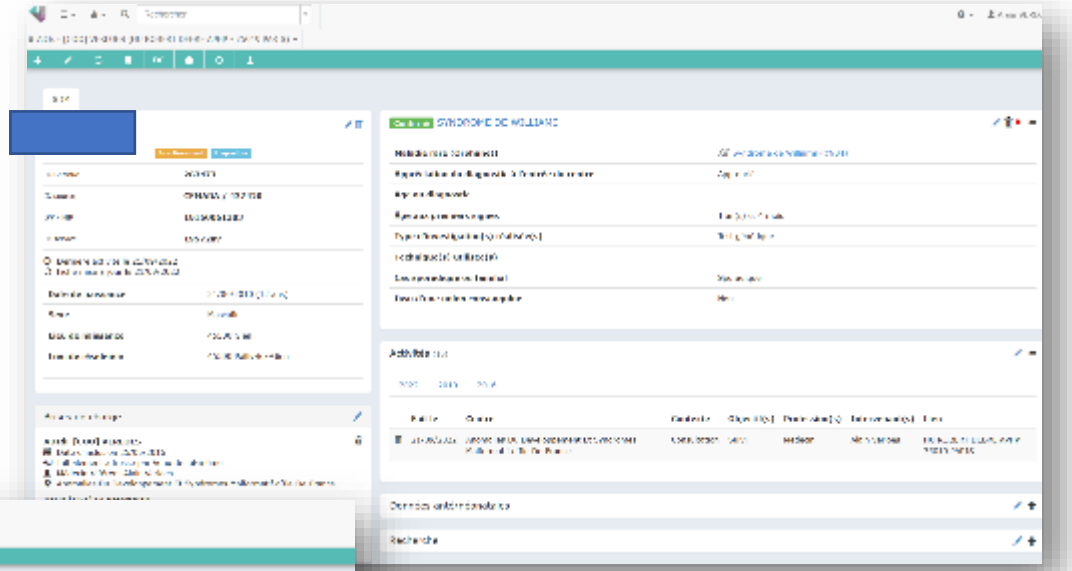
https://www.has-sante.fr/portail/jcms/c_1340879/fr/protocoles-nationaux-de-diagnostic-et-de-soins-pnds

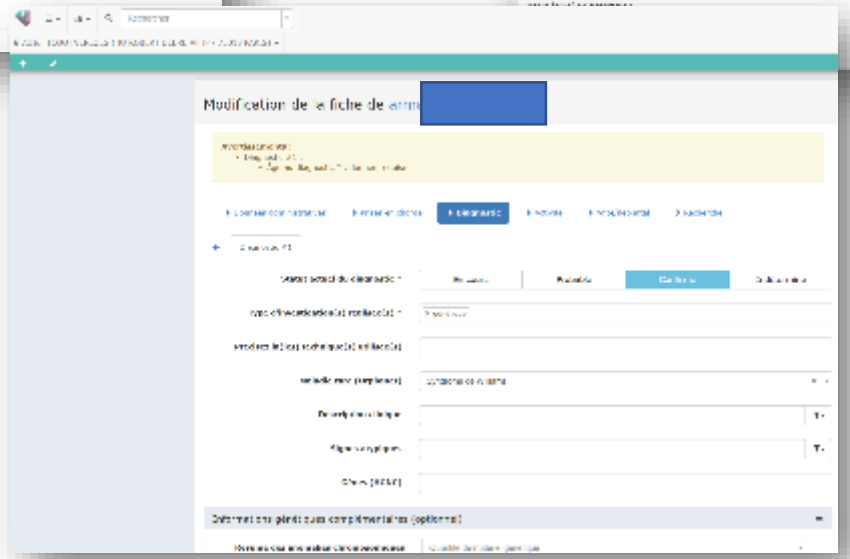
février 24

European Reference Networks ITHACA

BaMaRa & BNDMR: diagnosis & activity registry







- 1 single nat.l database for all CRMR
  - 1 identifiable DB/hospital (BaMaRa)
  - → 1 unique anomyzed Heath Data Warehouse (BNDMR)
- More than 1.000.000 patients entered since 2007
- IT partitioning by site and CRMR
- Incorporated in IT system of most academic hospitals

European Reference Networks ITHACA

Filières vs ERNs

Liberté • Égalité • Fraternité
RÉPUBLIQUE FRANÇAISE
MINISTÈRE DES AFFAIRES SOCIALES ET DE LA SANTÉ

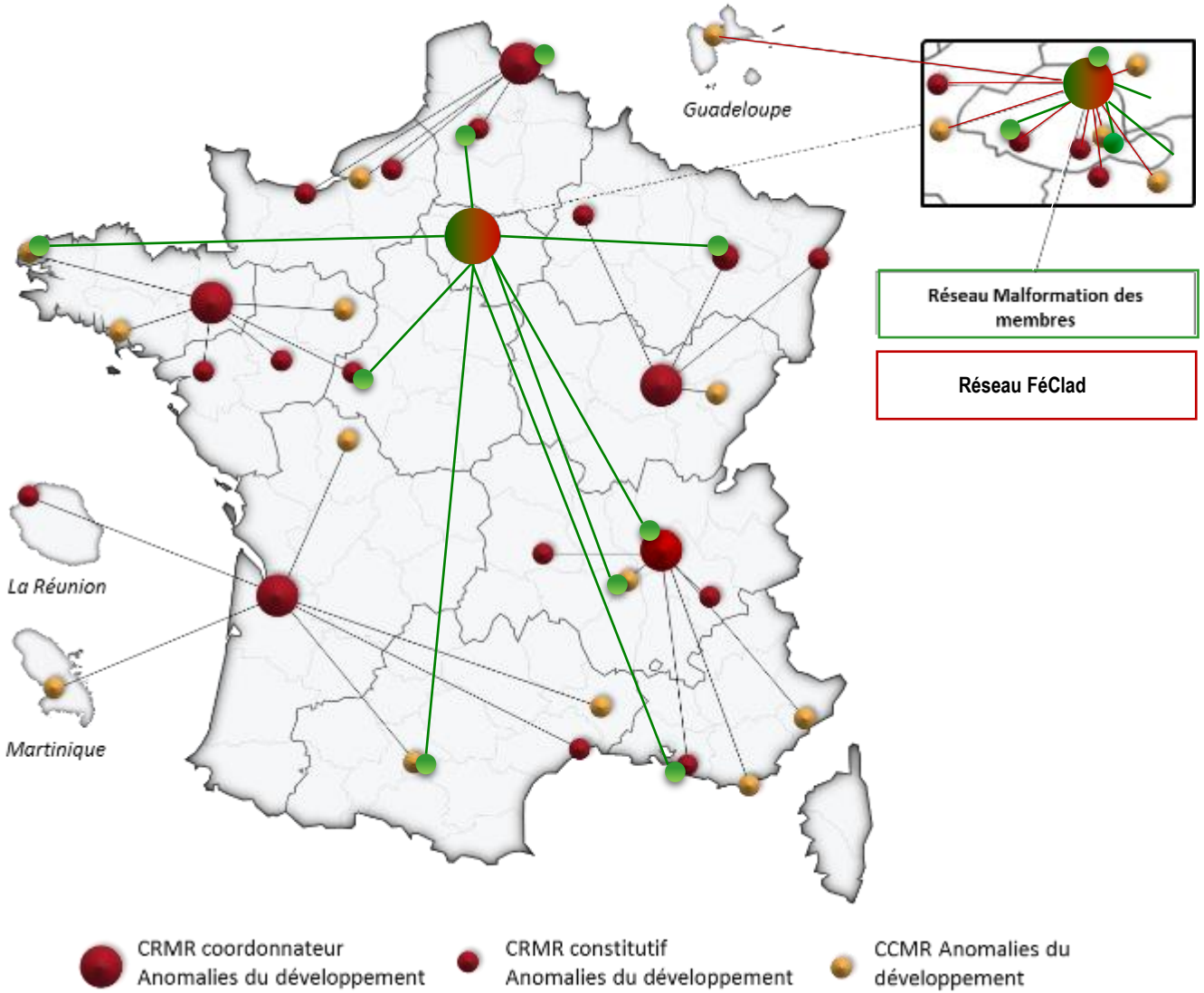
DIRECTION GÉNÉRALE DE L'OFFRE DE SOINS

CORRESPONDANCES FSMR/ ERN

	ERN	FSMR
1	**Craniofacial Anomalies and ENT Disorders** (Congenital anomalies of the skull and face, including hearing disorders and upper airway anomalies)	TETE COU - SENSGEN
2	**ENDO ERN** (Rare Endocrine Diseases)	FIRENDO
3	**EpiCARE** (Rare and Complex Epilepsies)	DEFISCIENCE
4	**ERN EYE** (Rare Eye Diseases)	**SENSGEN**
5	**ERN SKIN** (Rare and Undiagnosed Skin Disorders)	**FIMARAD**
6	**ERNICA** (Rare inherited and congenital digestive disorders, including rare gastrointestinal diseases)	FIMATHO
7	**eUROGEN** (European Reference Network in Rare and Complex Uro-recto-genital Diseases and Conditions)	NEUROSPHINX
8	**EuroBloodNet** (Rare Haematological Diseases, y compris maladies malignes)	**MARIH** – MCGRE- MHEMO
9	**ITHACA** (Intellectual disability TeleHealth And Congenital Anomalies) Rare Congenital Malformations and Developmental Anomalies and Rare Intellectual Disability	ANDDI - DEFISCIENCE
10	**ERN LUNG** (Rare Respiratory Diseases)	RESPIFIL - MUCO/CFTR
11	**MetabERN** (Rare hereditary metabolic disorders)	G2M
12	**ERN RND** (Rare Neurological Diseases)	BRAIN TEAM
13	**ERN BOND** (Rare Bone Diseases)	OSCAR
14	**GUARD-HEART** (Rare diseases of the heart)	CARDIOGEN
15	**EURO-NMD** (Rare Neuromuscular Diseases)	FILNEMUS FILSLAN
16	**RITA** (Rare Immunodeficiency, Autoinflammatory and Autoimmune Network)	FAIIR
17	**RECONNET** (Rare Connective and Muscoloskeletal Dieases)	FAIIR
18	**RARE-LIVER** (Rare hepatological diseases)	FILFOIE
19	**VASCern** (Rare Multisystemic Vascular Diseases)	FAVA-Multi
20	**ERKN** (Rare kidney diseases)	ORKID
21	***TRANSCHILD*** *(Transplantation in children incl. HSCT, heart, kidney, liver, intestinal, lung and multiorgans)*	Aucune
22	***GENITURIS*** *(Genetic Tumour Risk Syndromes)*	Aucune
23	***PaedCan ERN*** *(European Reference Network on Paediatric Cancer)*	Aucune
24	***EUROCAN*** *(Rare Adult Cancers)*	Aucune

Reference Networks ITHACA

Le réseau de soin AnDDI-Rares



filière de santé maladies rares

Réseaux AnDDI-Rares 2023

AnDDI-Rares

36 CRMR + 57 CCMR = 93 centres

- AD: **24 CRMR + 21 CCMR = 45 centres** *(+9 sites)*
- CeRefAM: **3 CRMR + 12 CCMR = 15 centres** *(+1 site)*
- GenoPsy: **5 CRMR + 6 CCMR = 11 centres** *(nouveau)*
- Spin@: **3 CRMR + 10 CCMR = 13 centres** *(nouveau)*
- PaRaDiGM: **1 CRMR + 8 CCMR = 9 centres** *(nouveau)*

+169%

génopsy

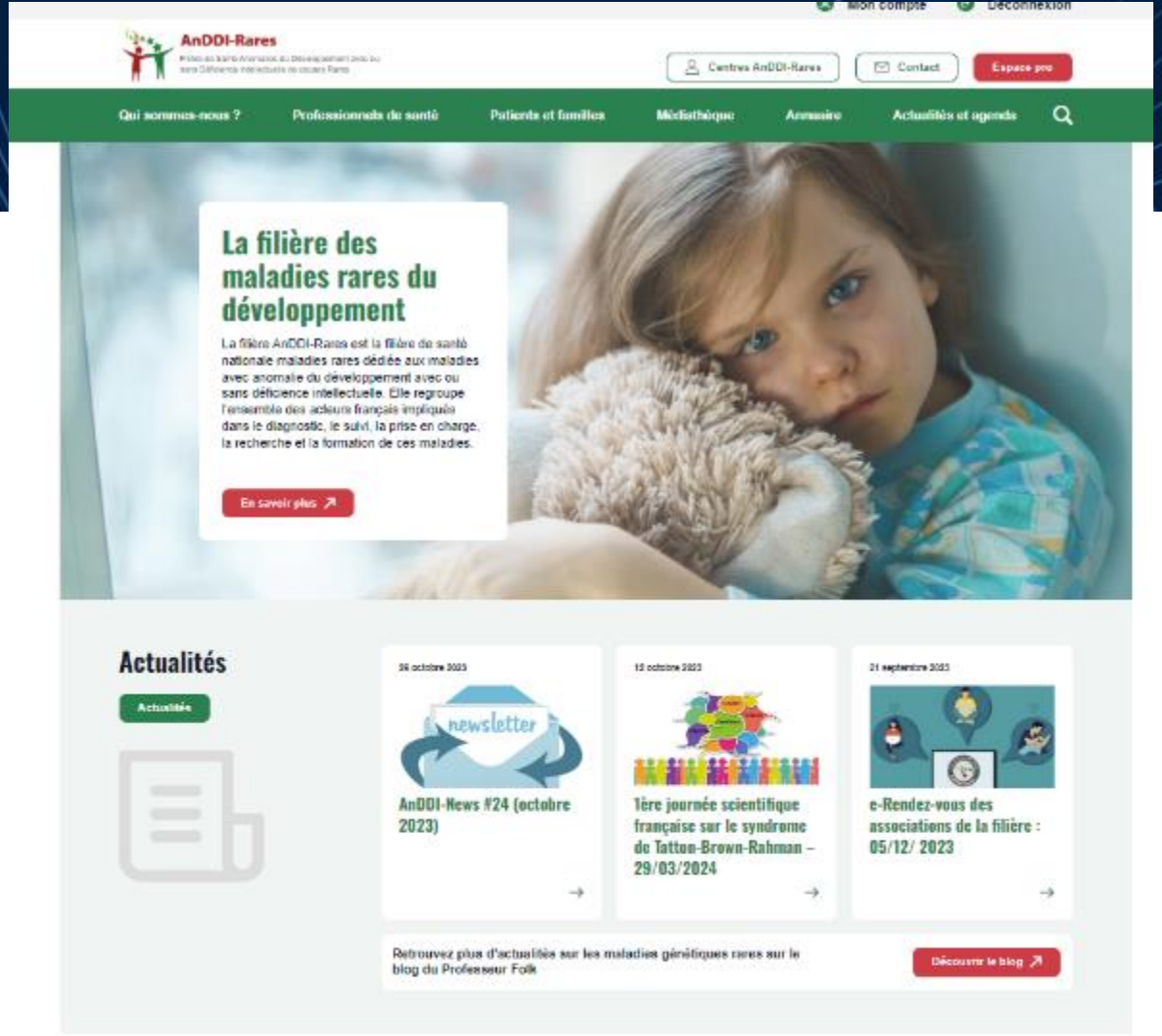
spin@ ANOMALIES VERTÉBRALES ET SPINA BIFIDA

CoPil – 21 septembre 2023 7

European Reference Networks ITHACA

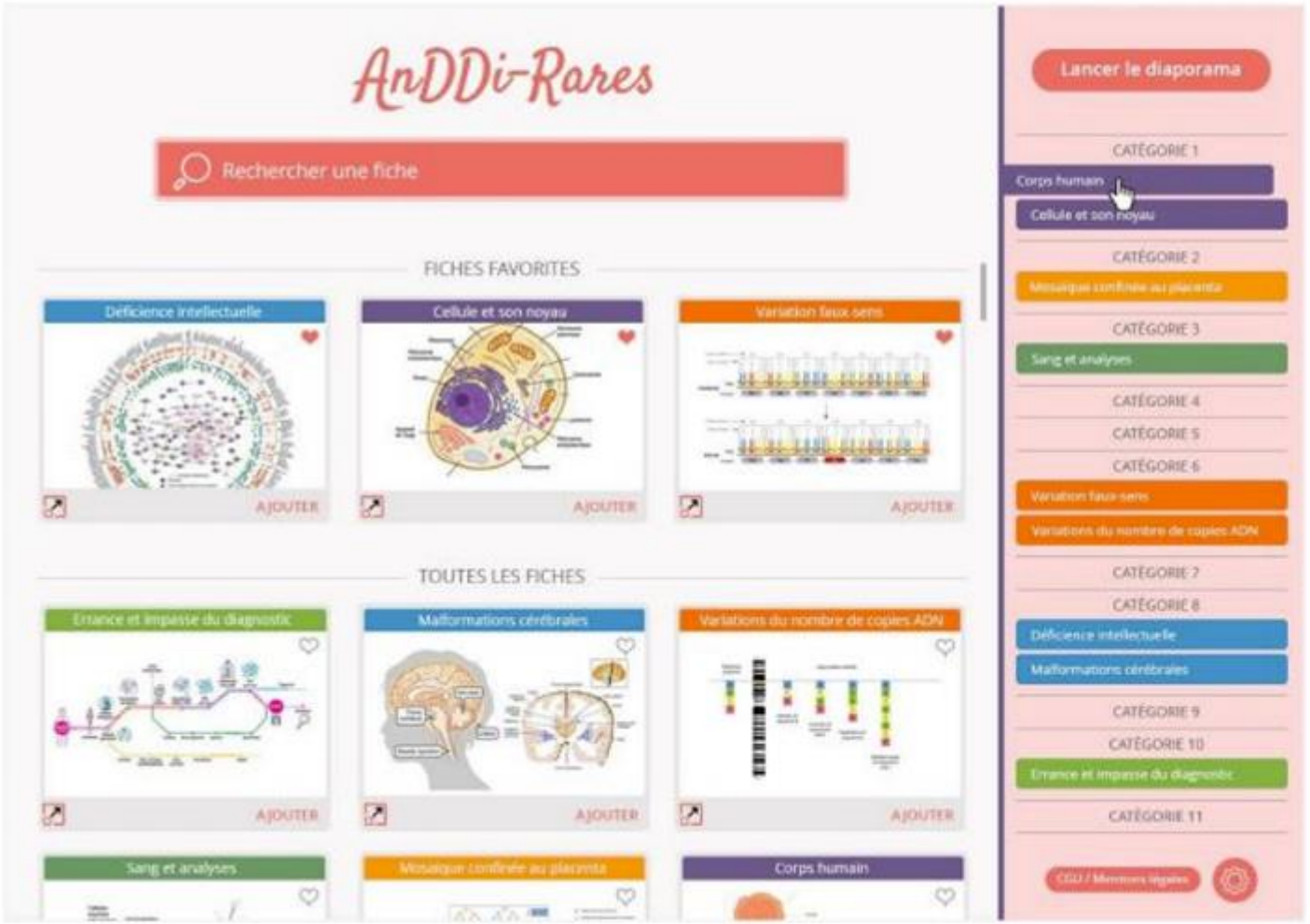
Site Web

- *Diagnostic et PEC*
- *Expertise pluridisciplinaire*
- *Communication*
- *Recommandations*
- *BNDMR*
- *Recherche*
- *Médico-social*
- *Formation*
- *Europe*
- *Associations*
- *Interfilière*
- *SoFFoet*



février 24

Axe 1 – AnDDI-Clic



Collaboration between ITHACA/French subsidiaries and African countries

- On the Web
  - Webinars
  - Free downloadable teaching material
  - ITHACA Calls for Collaboration
  - Treatment Protocols
    - French sectors
    - European sectors
  - MOOC BIG
  - Access to French inter-university diplomas supported by the course (dysmorphology, personalized medicine, etc.)
  - EuroDysmorpho
- Co-development?
  - Francophone : AnDDI-Rares / ITHACA (as an EU-Africa collaboration)
  - English Speaking : ITHACA

Thank you for your attention !