SPANISH PRESIDENCY COUNCIL OF THE EUROPEAN UNION

European Reference Networks

Training on rare diseases : the ERN experience and vision for the future

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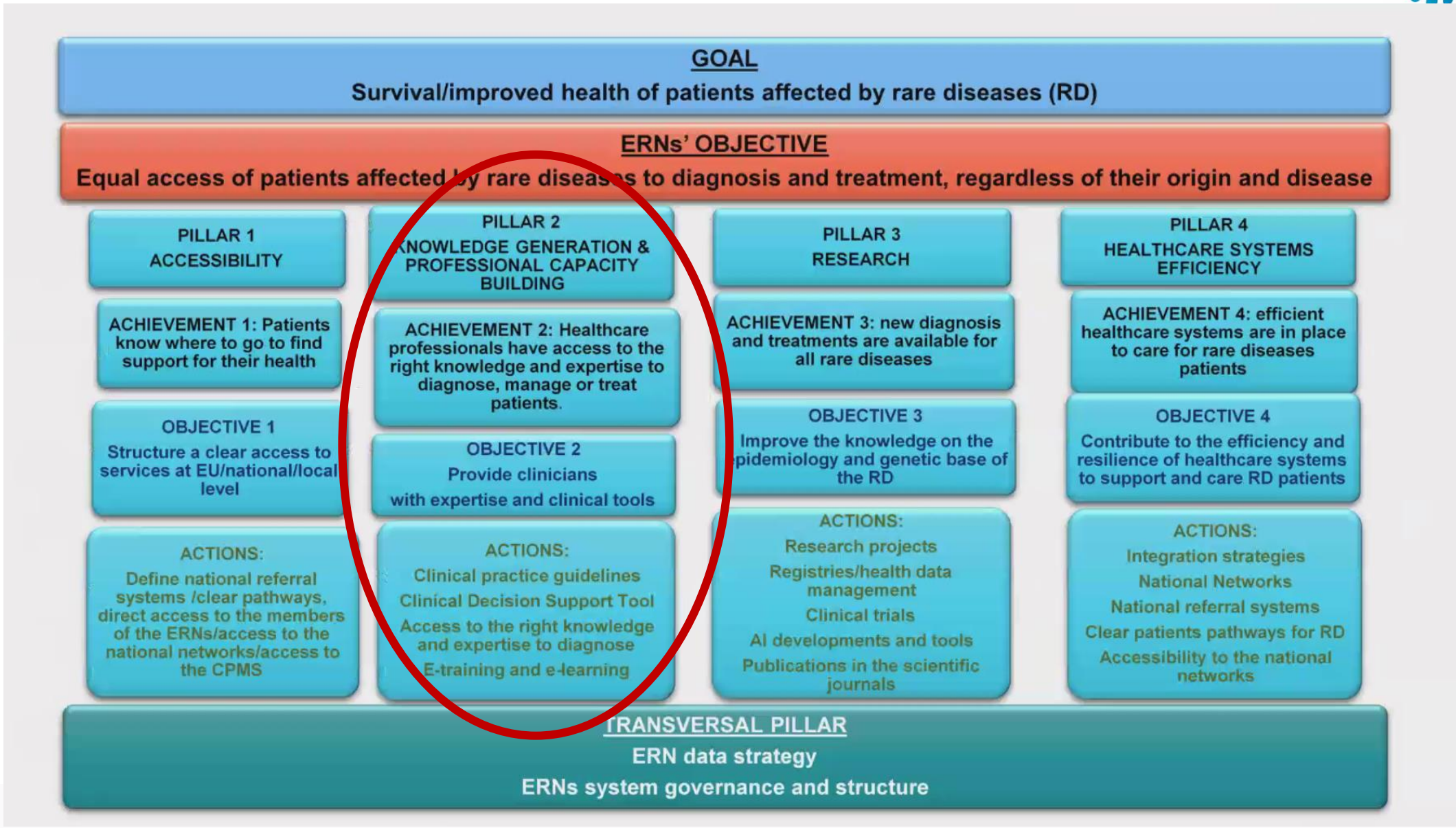
Chair of the Knowledge Generation Working Group

THANKS TO ALL THE ERN COLLEAGUES CONTRIBUTING TO THIS PRESENTATION

European Reference Networks



The ERN Strategy on Training and Education on Rare Diseases and Complex Conditions

To propose a joint strategy (MS, ERNs,stakeholders) for training and education to **overcome the gap** between the increasing awareness on rare and complex diseases and the lack of a specific training in the medical curricula.

Background principles:

- *ERNs are not Universities;*
- *ERNs are not expected to play the role of Scientific Societies;*
- *ERNs have a "patient care" mission*
- *Clinical Research is an obligation for clinician members of ERN medical teams, but training in research is also the mission of the EJP RD (close concertation)*

The curricula will be based on defined catalogues of training contents focusing on rare diseases or diagnostic tools and techniques as well as surgical techniques, which will be developed by the ERNs' educational working groups and approved by the Network boards.

The curricula should have a defined duration (to be defined by ERNs; not less than 1, not more than 3 years). Trainees should formally sign up for the curricula.

In addition, short- and medium-term exchange training fellowships (3 to 6 months duration) in ERN member centres with complementary expertise will be encouraged.

CME courses, training workshops, and summer/winter schools organized or validated by the ERN can be incorporated in the curriculum.

Registration should be open to postgraduate physicians from ERN member and Affiliated Partner centers who have completed a minimum level of specialization training (details to be defined by the ERNs).

The development of rare and complex disease curricula also for other healthcare professionals working in ERN teams (nurses, technicians) may be considered when appropriate.

All recognized training elements will be assigned by scores ('ERN training credits'). A minimum cumulative number of credits must be reached during the training program.

Measures to evaluate knowledge transfer should be incorporated (e.g. post-Webinar multiple choice questions, performance monitoring in eLearning courses, final online exam).

The curricula should combine periods of physical training in ERN centres with elements of online learning organized by the ERNs (such as Webinars and case-based e-Learning programs).

The Webinars and eLearning cases, focusing on rare diseases, should cumulatively cover all main items of the training catalogue during the training period. Provision by the EC of a - secure but readily ccessible - central online platform for data storage and e-learning case development would be welcome.

A certificate should be issued upon successful completion of the ERN training program. Recognition by European and national postgraduate education authorities should be aimed for.

In agreement with the ERNs' mission, no fees should be raised by the HCPs, members of ERNs, for the on-site training programs and all organizational costs should be covered by the central coordination budgets.

ERN ACADEMY

The ERN Academy is a tool which will facilitate the activity of the ERN Knowledge Generation activity

The ERN Academy will involve the following components:

- eTraining/eLearning Moodle-based environment and support to the content generators (the ERNs) to produce the deliverables and manage the system
- Repository-library of all the “products” of the ERNs (the ones included under the agreed taxonomy and any other produced by the ERNs) with a powerful search engine and structure
- Integrated web conferencing system for the training and learning activities
- A central IT and development support and helpdesk for all the activities and needs of the ERN Academy.

ERN ACTIVITIES ON EDUCATION

- Think tank: Subgroup “Training and Education” of the ERN Working group on Knowledge Generation
- Organization by ERN, based on field of expertise, connexions with National and EU Societies, and pre-existing achievements
- Learning & e-Learning
- Webinars
- MOOCS & Self training modules
- Workshops and symposium
- Formal courses
- Support for EU-wide trainings
- Training
- Short time exchanges
- Just a rapid visit of some ERNs… but hundreds of other initiatives !!!

WEBINARS

- Easy support for e-learning
- Synchronous & interactive… OR not
- Recordings available
- ZOOM, TEAMS…
- Advertisement through ERN NL and Organizing partners
- Usually opened to participants outside the Network
- Further dissemination through dedicated YouTube channels (some with > 100 videos)

- Targets
- Professionals : in English
- Lay people : language may be limiting

EDUCATION AND TRAINING PROGRAMME

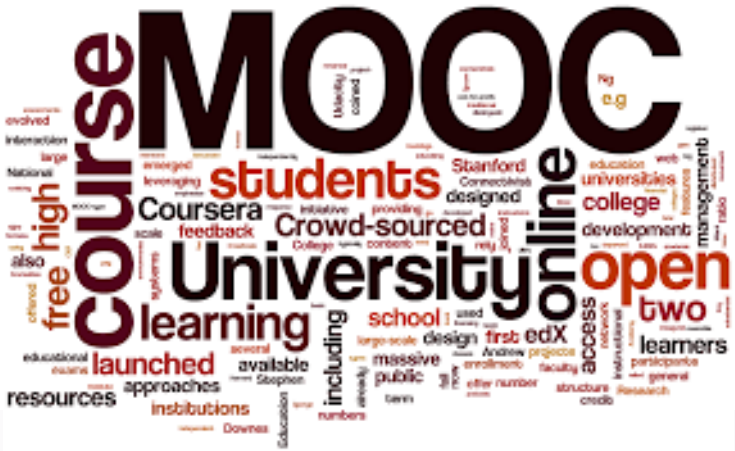
Type/Format	Event title	Date
Live Webinar	#01 Webinar: Gender issues in COVID-19 pandemia	22/06/2021
Live Webinar	#02 Webinar: EuRR-Bone: what and how?	28/06/2021
Live Webinar	#03 Webinar: Talking about FD/MAS	07/09/2021
Live Webinar	#04 Webinar: Talking about the type 2 collagen disorders	28/09/2021
Recorded webinar	#05 Webinar RD day 2021: COVID-19 and psychological impact – Italian w/ English subtitles	22/02/2021
Recorded webinar	#06 Webinar RD day 2021: COVID-19 and vaccines – Italian w/ English subtitles	28/01/2021
Live Webinar	#07 Webinar Human Body Movement & Scan Analyses: in state-of-the-art laboratories, in patients' local area, at home	24/03/2022
Recorded webinar	#08 ERN BOND, the last five years and the future from the "5th Annual meeting of clinical and molecular diagnosis of skeletal dysplasias: from research to clinical practice"	20/04/2022
Live Webinar	#09-#10 ECTS-ERN BOND Webinar Series Rare Bone Diseases: Melorheostosis - First and second part	07/02/2023; 14/03/2023
Webinar	#11 Webinar RD day 2021: Tools for remotely following up patients with rare skeletal diseases – Italian w/ English subtitles	04/03/2021 (live); 21/03/2023 (recorded)
Webinar	#12 Webinar RD day 2021: COVID-19 and indications for patients with OI – Italian w/ English subtitles	21/01/2021 (live); 21/03/2023 (recorded)
Recorded webinar	#13 Musculoskeletal avatar. A possible solution for creating digital twins in children with skeletal dysplasia	31/03/2023
Webinar	#14 Webinar RD day 2021: Biological material in scientific research – Italian w/ English subtitles	18/03/2021 (live); 01/04/2023 (recorded)
Live Webinar	#15 ECTS-ERN BOND Webinar Series Rare Bone Diseases: The pathogenesis and consequences of having too much bone	31/05/2023

Courtesy of Luca Sangiorgi, ERN Bond

MOOCS, ON LINE ENCYCLOPEDIAS AND OTHER SELF-LEARNING TOOLS

- Many initiatives as asynchronous or synchronous (Massive Open Online Courses, MOOCS) e-learning tools
- Supported initally by a specific CEF funding

- From focused information (« pills of knowledges ») to large e-textbooks
- MetabERN : encyclopedia en metabolic diseases (coming 2024)
- ITHACA: APOGeE
- Many opp\ortunities for trans-ERN developments



European Reference Network
for rare or low prevalence complex diseases
Network Vascular Diseases (VASCERN)

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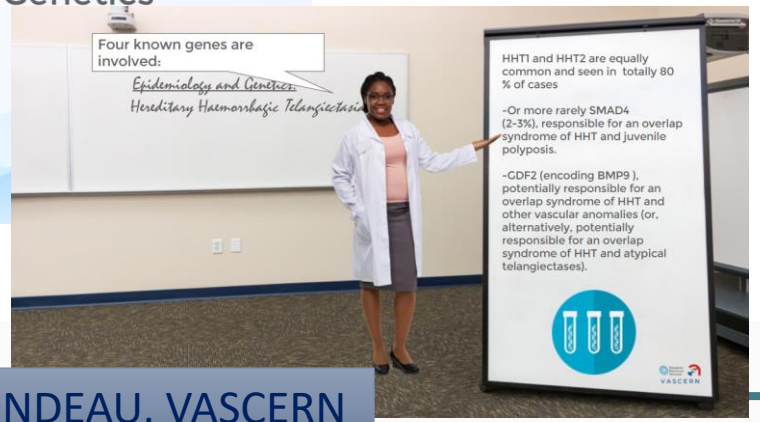
- Pills of Knowledge videos on several topics to increase knowledge of rare vascular diseases.

- E-Learning modules on different rare vascular diseases.

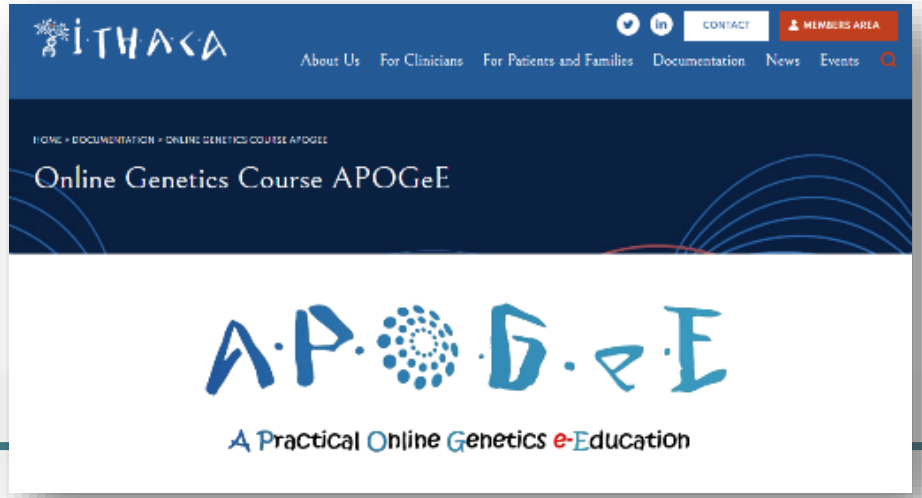


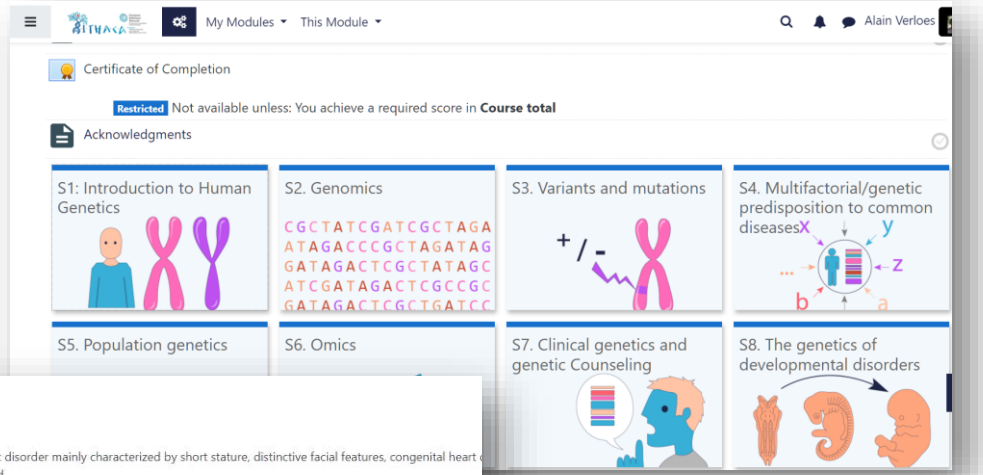
- VASCA Magazine by the VASCERN Vascular Anomalies Working Group.
- A true collaboration between clinicians and patients.

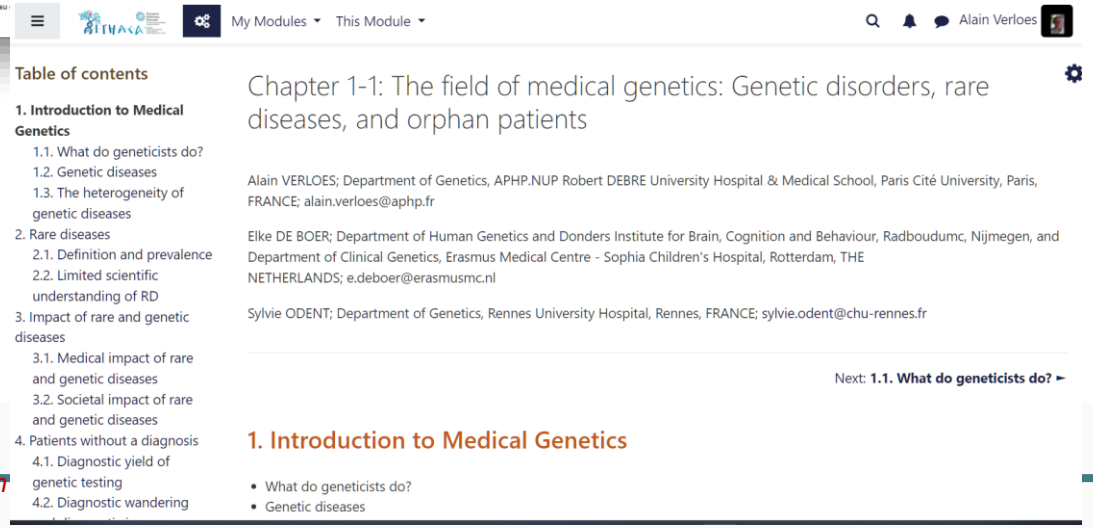
Courtesy of Guillaume JONDEAU, VASCERN

APOGeE (ITHACA)

- APOGeE
- Online e-handbook on medical genetics
- 100 contributors
- Adopted as reference for ECCMG certificate for Genetics (UEMS)
- With strong accent on NDD and developmental anomalies
- In development (beta version)
- Achievement T4, 2023
- Others: 2 MOOCs, 1 serious game…







FORMAL COURSES

- Several ERNs have formal courses or contribute/support existing courses

- Postgraduate ± accredited training and annual “summer school” (EURACAN) or “winter school” (for Fetal Pathology: ITHACA)

- Patient-derived eLearning formats for the training of students and residents (MetabERN).

ERN-RND POSTGRADUATE CURRICULUM *PROJECT*

Inter-ERN initiative
RND, EURO-NMD, EpiCARE

EAN, EPNS,
EuroNMD, EpiCARE

BoK
(Body of Knowledge)

TRAINING CONTENTS

1. **Webinars: convey knowledge**
2. **Educational cases (eLearning): apply knowledge**
3. **Stays at expert centres: consolidate practical knowledge**

PARTICIPATION MODALITIES

1. **Eligibility and participation criteria**
2. **Technical administration**
3. **Software/Platform/Learning Managamenet System**
4. **Assessment / Certificate qualifying criteria: time and success rates**

Module per DG

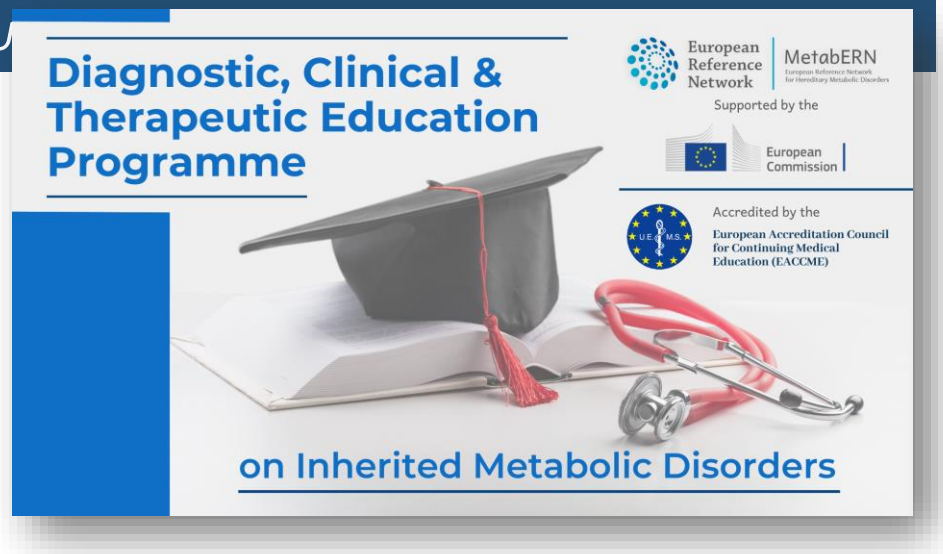
ERN-RND

European clinicians

TOPICS

1. Neurogenetics
2. Neuroimaging
3. Clinical research
4. Multidisciplinary care & Neurorehabilitation
5. Digital Care/Telemedicine
6. Patient perspective

Courtesy of Holm GRAESSNER, ERN RND



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The target audience is represented by **post-graduate medical students, medical residents, general practitioners**, and **medical specialists.**

Currently, the course consists of **11 modules**, each including video lectures by experts in the field of IMDs, chosen among MetabERN members. It is designed to be continuously updated by adding new modules.

The course has been accredited by the **European Accreditation Council for Continuing Medical Education (EACCME)**, an institution of the European Union of Medical Specialists (UEMS).

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TREAT-NMD Neuromuscular Network

European Reference Network for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

5th Neuromuscular Translational Summer School

July 10-14, 2023

Leiden University Medical Center, the Netherlands

Under auspices of EURO-NMD and TREAT-NMD

European Reference Network | Neuromuscular Diseases (ERN EURO-NMD)

Neuropathies Rehabilitation Summer School

25 May 2023

Do you want to improve your knowledge on current and future modalities of rehabilitation in rare and complex peripheral neuropathies ?

Join us at the Summer School on Neuropathies Rehabilitation in September 2023 in Rome 🇮🇹 !

Courtesy of Teresinha EVANGELISTA, ERN EURO-RMD

Preceptorship structure

- Participants hosted in an expert HCPs (and selected via an open call)
- Intense training from 2 to 10 days
- From 1 to 5 participants
- Awarded with CME points

- Train hematologist in patient care
- To learn or improve knowledge in clinical research
- To stimulate cross-border collaborations
- To facilitate the Harmonization of specialties curricula in Europe

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EBAH CME ACCREDITED

EuroBloodNet

Accreditation of CME Point via assessment of an European independent accreditation body of Hematology

Submission to the program to EBAH as **formal recognition** of the delivery of **high quality educational programs**

NB: → Direct tutorial activity in laboratory and/or clinical consultation

Courtesy of Pierre FENEAUX, EuroBloodNet

Co-funded by the European Union

With collaborations between ePAGs and local EpiCARE members we started translating our leaflets in several languages

European Reference Networks
EpiCARE

Rare epilepsies leaflets

Patient and caregivers leaflets are developed to give precise and accessible informations on rare and complex epilepsies. With **one part for healthcare profesionnals, and one part for patients and their families or carers**, these documents detail comprehensively what to expect when facing a rare epilepsy, and how to manage care.

Unless mentioned otherwise, all leaflets are in english. We are working on translating them in as many languages as possible with the help of patients associations all over Europe, so check back regularly!

Patients & caregiver leaflets

European Patient Advocacy Group

Read and download the following leaflets:

- **Dravet** Syndrome leaflet (EN) / Dravet versione italiana (IT) / Dravet Versiunea română (RO) / Dravet hrvatska verzija (HR) / Dravet deutsche Version / Dravet Norsk versjon
- **Hypothalamic Hamartoma** leaflet / Hypothalamic Hamartoma Versiunea română (RO) / Hypothalamic Hamartoma Hrvatska verzija (HR)
- **Ring Chromosome 20** leaflet / Ring Chromosome 20 Hrvatska verzija (HR)
- **Alternating Hemiplegia of Childhood** leaflet (EN) / AHC Hrvatska verzija (HR) / AHC Versione italiana (IT)
- **GLUT1 Deficiency Syndrome** leaflet (EN) / GLUT1 Hrvatska verzija (HR) / GLUT1 Versiunea română (RO)
- **CDKL5 Deficiency Disorder** leaflet (EN) / CDKL5 Versión española (ES) / CDKL5 Versão portuguesa (PO) / CDKL5 Hrvatska verzija (HR) / CDKL5 Versiunea română (RO)

And a new initiative:

Considering the specificities of the epilepsy monitoring activities we created a Work Group for Nurses and EEG technicians, to share practices and develop a curriculum.

First workshop at the Utrecht annual meting:
76 participants from 24 countries !!!

The 3rd edition of an annual workshop, held in Rome to treat 2 major topics in the field
+ one session reserved to early career epileptologists
+ another one to Pharma
(treatment innovations)

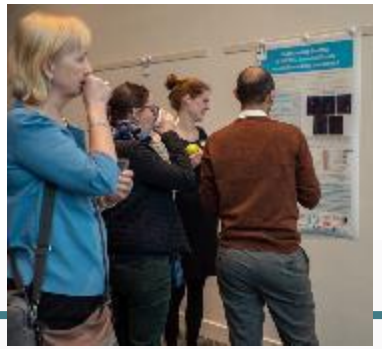
Courtesy of Alexis ARZIMANOGLOU, EpiCare

WORKSHOPS AND SYMPOSIA

- ERN system allows the organisation of Workshops
- Private co-funding not allowed
- Support for catering and/or invited speakers

- Learning/training days connected to Annual Board and other ERN events

1st EuroNDD WORKSHOP



- Objective: crossed vision across specialties on **NEURODEVELOPMENTAL DISEASES**
- Mixing in a unique event Neuroscience, Genetics, Social Sciences and Medicine interested by ID, ASD, and other NDDs
- Promote networking across EU, Incubate projects, researchs…
- Invited speakers, oral presentation & posters
- EuroNDD 2023
- 240 participants (free meeting, catering supported by ITHACA)
- 3 half days
- Organistion : Prof. Tjitske Kleefstra (Rotterdam), Prof. Christiane Zweier (Bern)
- Organising Committee 2023: Dr Laura de Graaff-Herder, Dr Marco Tartiglia, Pr Zeynep Tümer
- EuroNMD 2: Lisbonne 04/24 // EuroNDD 3: Poland ?

WORKSHOP ON TRANSITION (ERN RARE-LIVER)

- Face-to-face networking event involving patient teenagers (Ghent 2023)
- Aim: develop plan of action to improve age-appropriate and transitional care
- Starting from “a best model of practice” : to define the minimal but absolute requirements for transitional care.
- 15 adolescent Patients have formed the core group of the new build Youth panel that was also across ERNs. (TransplantChild and EJP RD)
- Organization interERN
- Three subgroups : communication, organization, and education → identification of actions to tackle in 2023-2024
- Several FU meetings foreseen, ending with publications

Courtesy of Sabrina Bernhardt, ERN RARE-LIVER

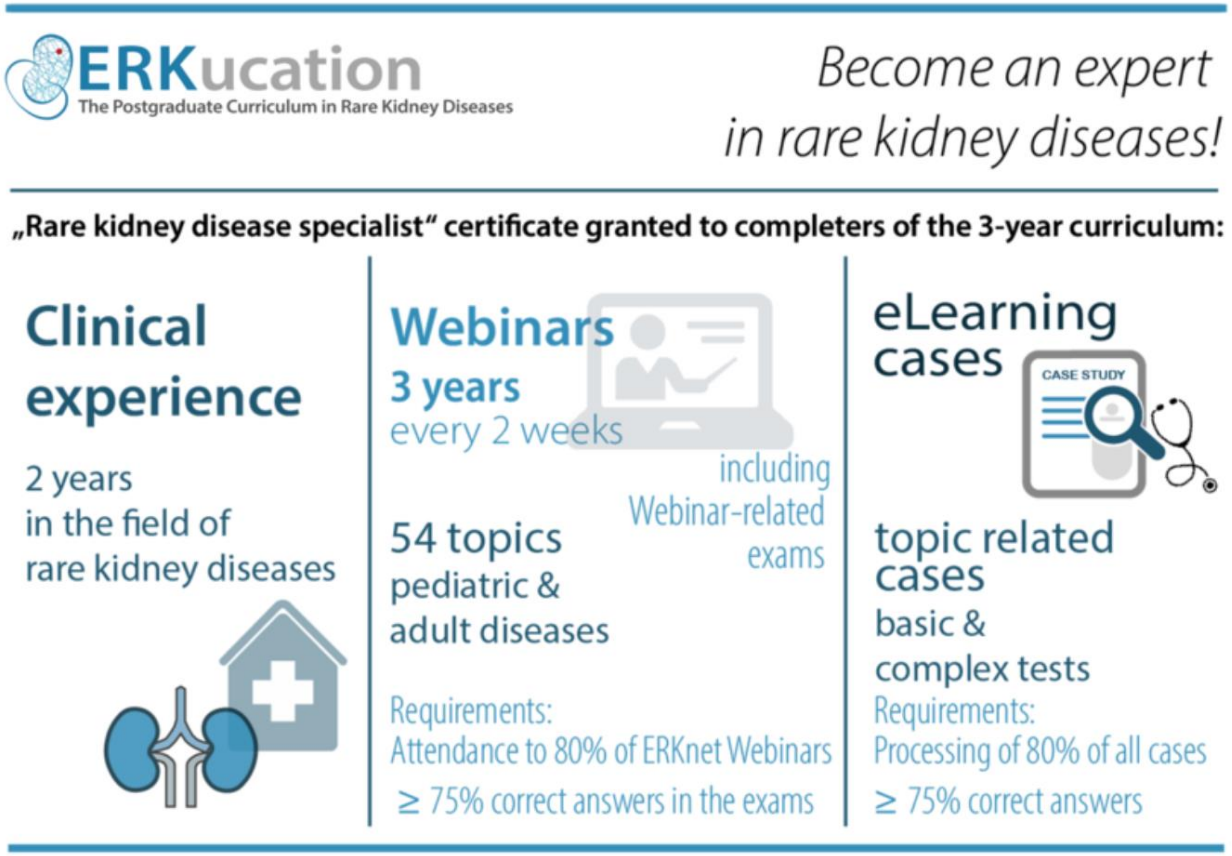
FORMAL COURSES AND EU CERTIFICATES/DIPLOMA

- Organisation or support/co-organization of formal Specialized courses

- Support to EU certificates
- ITHACA: supports of the EUMS Certificate in Genetics

- Co-organisation with EU Societies

ERKNET POSTGRADUATE CURRICULUM



The **ERKNet Postgraduate Curriculum** is a structured 3-year online programme consisting of webinars and case-based eLearning covering the whole spectrum of rare kidney diseases.
The webinars are presented twice a month by outstanding paediatric or adult nephrologists, nephropathologists and geneticists.

In total **347 interested ERKNet healthcare professionals from 65 health care providers in 22 European countries** enrolled to the ERKNet Postgraduate Curriculum in Rare Kidney Disease.

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ERN EXCHANGE PROGRAMME 2021-2022

- The exchange programme is meant to meet goals and strengthen capacity at the network level.
- The thematic scope includes medical practice and skills but also organisational aspects of a network.
- Each ERN defines the strategic goals and priorities of the exchange programme, according to the specific situation of the network.
- Health professionals working in the ERN centres are eligible for the programme – this includes all disciplines related to the relevant expertise area of the ERN.
- Formally affiliated member HCPs are also eligible for exchange visits.

THE GUIDELINE PROJECT

European Reference Networks

Consortium Members

COORDINATION	Fundación Pública Andaluza Progreso y Salud (**FPS**) Agencia de Calidad Sanitaria de Andalucía (**ACSA**)
PRODUCTION	Agencia de Evaluación de Tecnologías Sanitarias (**AETSA-FPS I+i**) Fundación Vasca de Innovación e Investigación Sanitarias (**BIOEF**) Instituto Aragonés de Ciencias de la Salud (**IACS**) Agència de Qualitat i Avaluació Sanitàries de Catalunya (**AQuAS**) Fundación Canaria Instituto de Investigación Sanitaria de Canarias (**FIISC**)
TRAINING	Escuela Andaluza de Salud Pública (**EASP**) Fundacio per la Universitat Oberta de Catalunya (**FUOC**)

Fundación Progreso y Salud
CONSEJERÍA DE SALUD Y FAMILIAS

Agencia de Calidad Sanitaria de Andalucía
CONSEJERÍA DE SALUD Y FAMILIAS

AETSA
Evaluación de Tecnologías Sanitarias de Andalucía

b+oef
berrikuntza + ikerketa + osasuna euskо fundazioa
fundación vasca de innovación e investigación sanitarias

IACS Instituto Aragonés de Ciencias de la Salud

Agència de Qualitat i Avaluació Sanitàries de Catalunya

FIISC

Escuela Andaluza de Salud Pública
CONSEJERÍA DE SALUD Y FAMILIAS

European Commission

European Reference Networks

Universitat Oberta de Catalunya

EMPOWERMENT OF PATIENTS

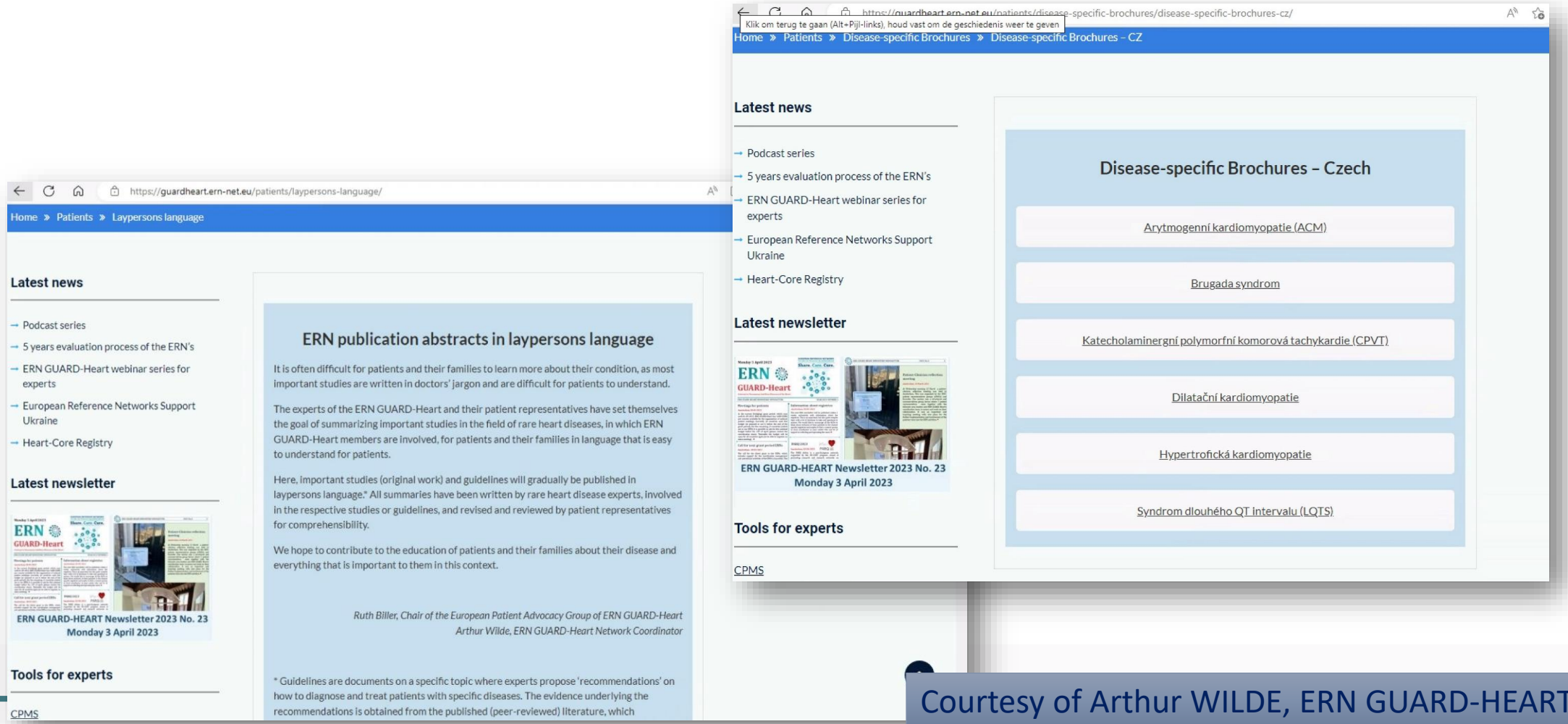
- Involvement of PAGs at many levels in all educatiuonal activities

- Provision of a simple language version of many documents (guidelines, publications, etc.)

LAYPERSON ABSTRACTS FOR PATIENTS AND TRANSLATED BROCHURES

European Reference Networks

- **Translation of all official ERN publications in layperson language (2022 / 2023)**



European Reference Network
for rare or low prevalence complex diseases
Network
Heart Diseases
(ERN GUARD-HEART)

Courtesy of Arthur WILDE, ERN GUARD-HEART

THE NEW CHALLENGES

- AWARENESS ON RD
- TEACHING RD AS PART OF THE CV IN MEDICAL SCHOOL
- NEW TECHNOLOGIES
- CREATION OF THE ERN ACADEMY
- CREATION OF THE ERN ENCYCLOPEDIA
- DATA GENERATION
- ARTIFICIAL INTELLIGENCE

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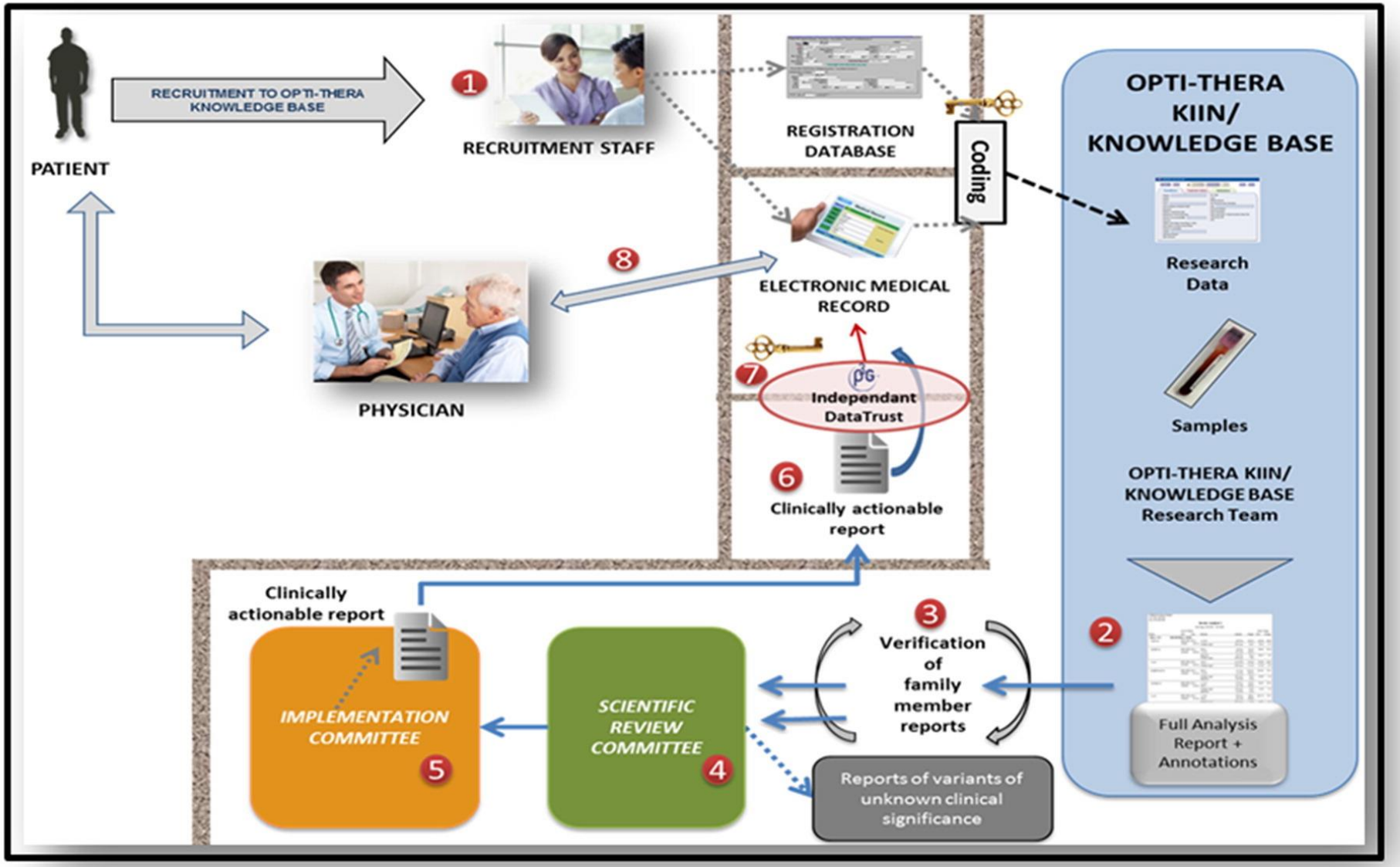


EUROPEAN REFERENCE NETWORKS

DISEASE ONTOLOGIES and THESAURUS

SOURCE: MeSH, MedDRA, Disease Ontology, ordo, mondo

EXCLUSION:
Neoplastic Disorders; Obsolete or NON-Rare; Other free terms like Cancers, melanoma,

EPIDEMIOLOGY

SOURCE: orphadata

DATA TYPE:
- Prevalence Class
- Epidemiology (descriptive)

SCIENTIFIC KNOWLEDGE

SOURCE: PubMed.gov

DATA TYPE:
- # of Publications

GENETIC KNOWLEDGE

SOURCE: OMIM

DATA TYPE:
- Gene details
- OMIM number
- OMIM group

CLINICAL KNOWLEDGE

SOURCE: U.S. National Library of Medicine ClinicalTrials.gov, Trialtrove

DATA TYPE:
- # of Clinical Trials

SYMPTOMS

SOURCE: hpo

DATA TYPE:
- # of Symptoms
- # of Organs affected

SoC AND FUTURE THERAPIES

SOURCE: orphadata, DRUGBANK

DATA TYPE:
- # of Approved Drugs
- # of Orphan Drug Designation Granted (EU and US)

RDs GENERAL KNOWLEDGE

SOURCE: orphadata

DATA TYPE:
- Disease Definition
- Classification
- Clinical description
- Etiology
- Diagnostic methods
- Differential diagnosis
- Antenatal diagnosis
- Genetic counseling
- Management and treatment
- Prognosis

PTS ASSOCIATIONS

SOURCE: orphadata

DATA TYPE:
- # of PTS Associations

EXPERT CENTERS

SOURCE: orphadata

DATA TYPE:
- # of Expert Centers

LABS & DIAGNOSTICS

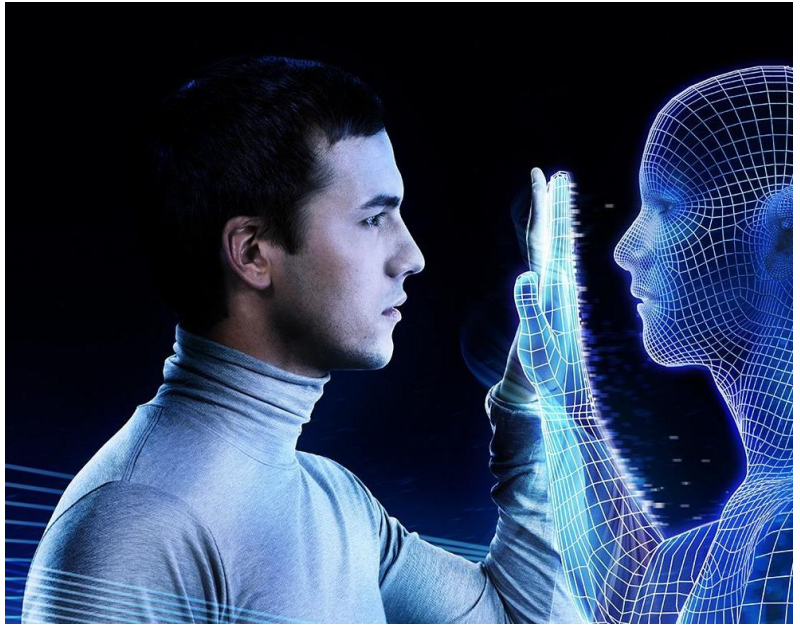
SOURCE: orphadata

DATA TYPE:
- # of Labs
- # of Diagnostics

BIOBANKS & REGISTRIES

SOURCE: orphadata

DATA TYPE:
- # of Biobanks
- # of Registries



THE DIGITAL TWIN PROJECT

SOURCE DATABASES: PubMed.gov, OMIM, hpo, orphadata, Trialtrove, U.S. National Library of Medicine ClinicalTrials.gov, DRUGBANK

DATA PREPARATION

Open for Innovation KNIME

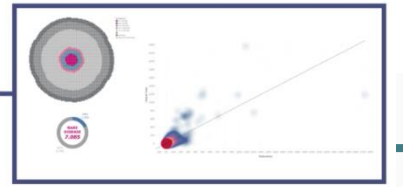
DATA ANALYSIS

Linguamatics

DATA VISUALIZATION

tableau SOFTWARE

VISUAL ANALYITICS



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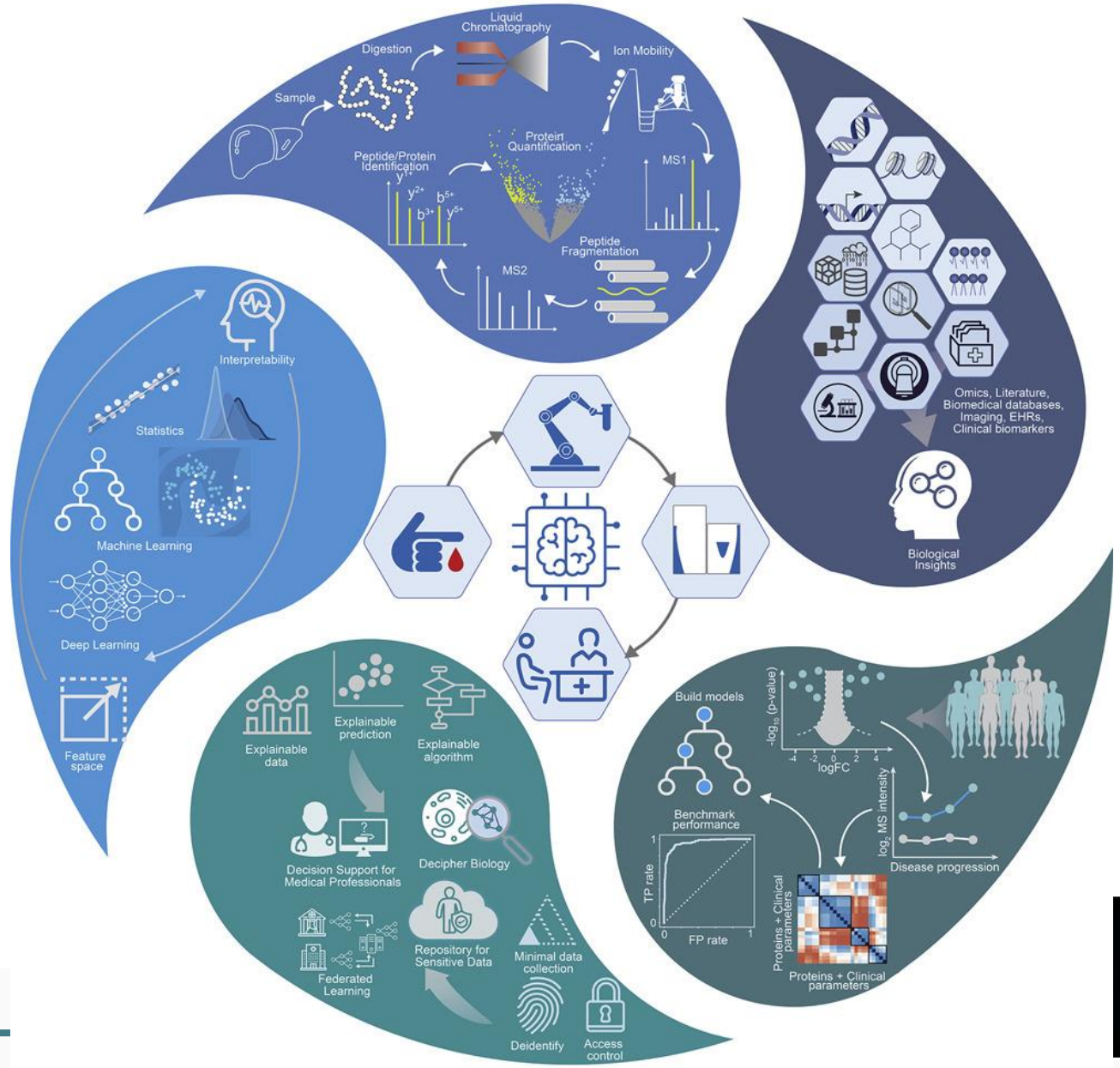
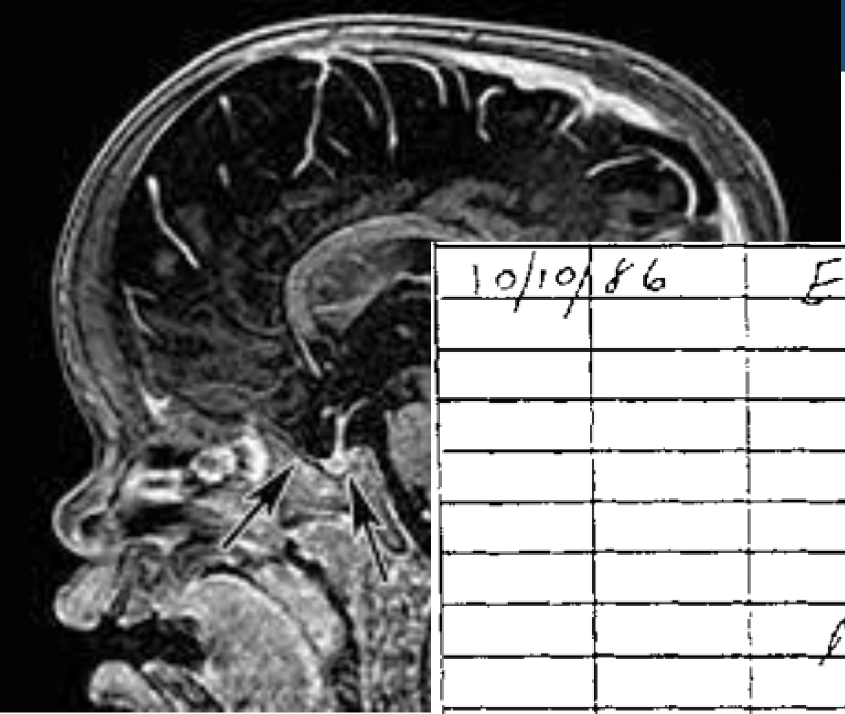


Mann M, et al. Cell Syst. 2021 Aug 18;12(8):759-770.



Blood test data	AUC	Optimal cut-off value	Univariate analysis: Number of patients		30-day survival (%)	p	Multivariate analysis: Odds ratio for 30-day mortality	95%CI	p
C-reactive protein	0.6752	5.4 mg/dL	≧5.4	385	29.4%	<0.0001	1.86	1.30 – 2.67	0.0007
			<5.4	376	55.5%		1		
Albumin	0.6275	2.8 g/dL	≧2.8	294	56.0%	<0.0001	1		0.0006
			<2.8	467	33.7%		1.9	1.31 – 2.76	
Total bilirubin	0.5909	1.3 mg/dL	≧1.3	151	23.8%	<0.0001	1.55	0.93 – 2.62	0.0953
			<1.3	610	46.8%		1		
Aspartate aminotransferase (AST)	0.5783	47 IU/L	≧47	218	47.3%	<0.0001			

SUMMARY

As a Clinical Data Analyst over 5 years of experienced in providing over view of data- base, contributing data management activities and tracking quality of data.

SKILLS

Process Improvement, Data Collection, Data Management Activities.

CE

yst

2011 – April 2016

nd communicated quality and utilization metrics for the hospital, partment chairs, and physician faculty to educate and align medical itor, and governmental guidelines.
ospitalists, specialists, and residents emphasizing action steps to bridge

rts to solve priority performance issues across silos.
hips with clinicians, care management, administrators, and vendor ccess.
takeaways, benchmarking Case Mix Index, Length of Stay, y, Costs, and Denials.
gement project activities according to Training Manuals and SOPs.
team meetings and partnered with the Clinical Research Associates to bject data and other project-related issues.

yst

2011

nsion of duties listed below at Pfizer Inc., GR&D

10/10/86 ENT:

Pt c Aspergillosis by path
Tolerating Amphotericin B well
Except for Nausea c poor po intake
Cr 1.7
Pt c retro headache & visual blurring c prednisone
PE: EOMI, PERRLA
HEENT: TM's clear & intact
Nose: ⊝ lesion
⊝ bleed
HP/NP: ⊝ ⊝ slight frontal sinus tenderness
LX – clear TVC mobile
will get head scan, neurology consult
Tx: stable
Plan: – Continue Amphotericin B
– plan as per

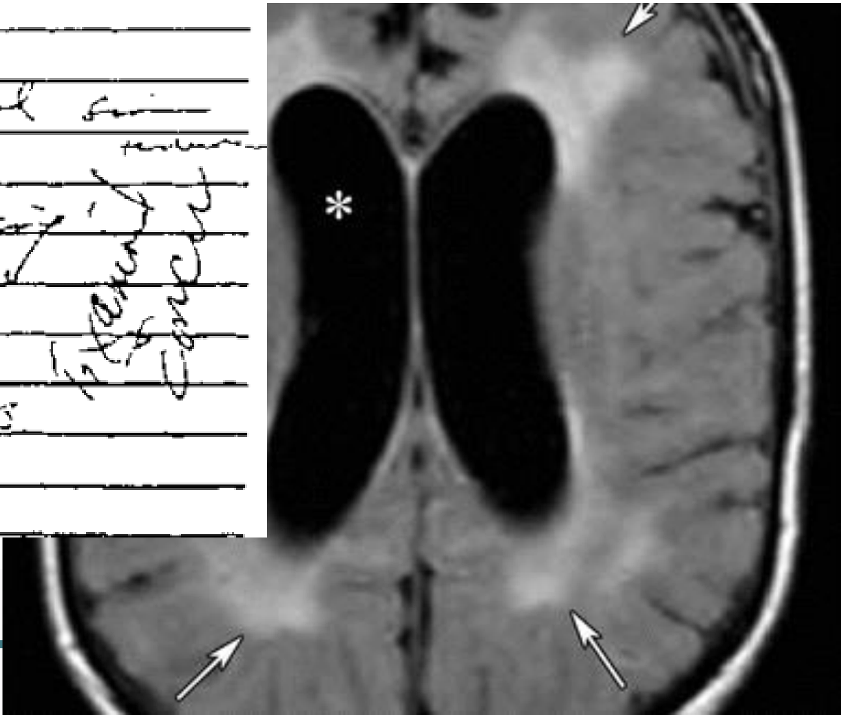
LE RADIOLOGY CONSULTING
entification

Card Number: 2435/98
Age: 45
Sex: Male
Maritial Status: Married
Occupation: Merchant
Address: Yirgalem

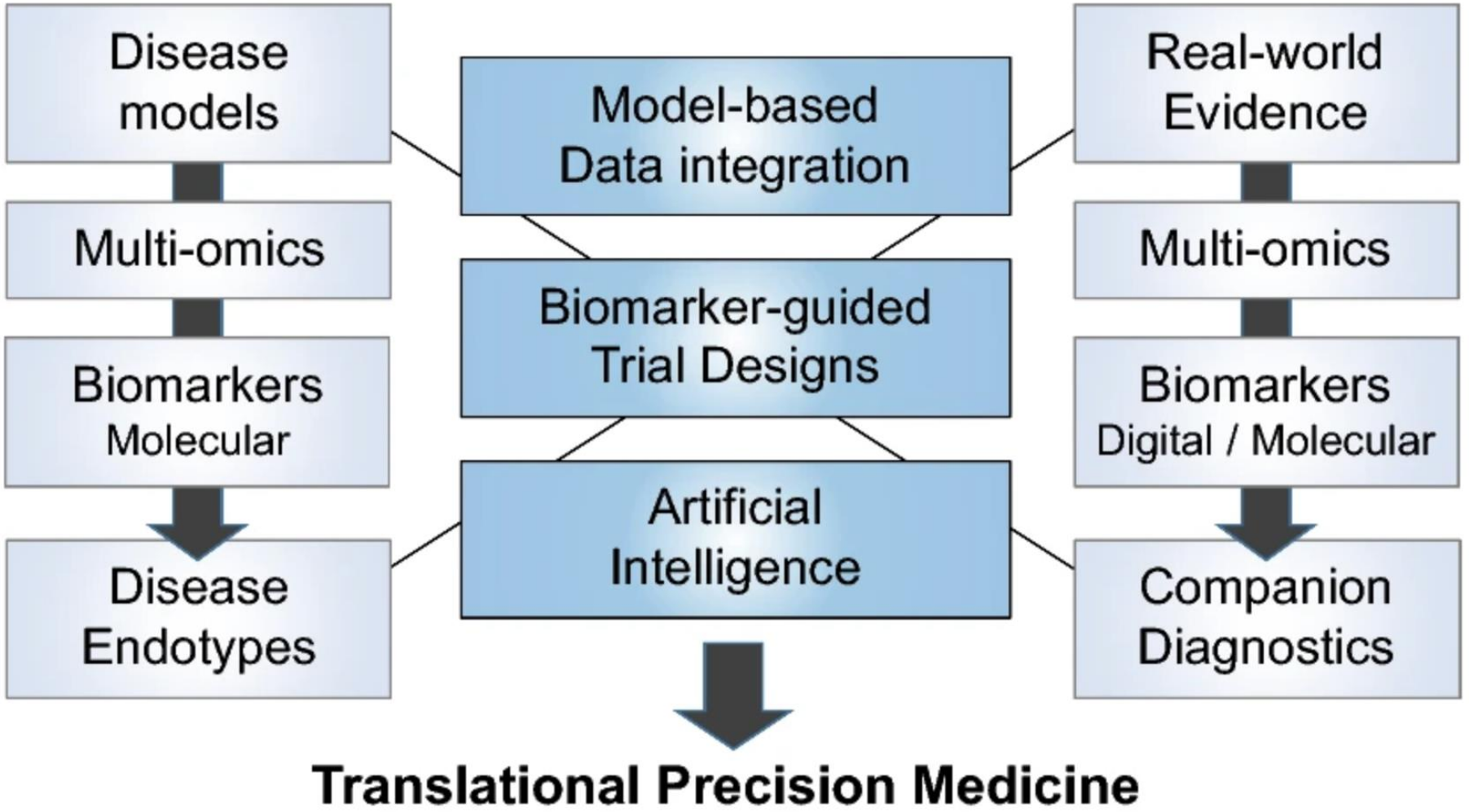
tient Clinical History
cough,fever and night sweating of 02 months. Hx of signific
eatment (if given)
antibiotics and analgesic.
levant Lab.Information
ESR 100mm/hr
Bc 7500
50%
40%
=05%
05%
evious related imaging findings
no
nsulting Physician Impression
Pulmonary TB R/O Bronchogenic Carcinoma
ffering Physician:Dr Zelalem Assefa
te of Consultation :09/03/06

Fasting insulin (μU/mL)	11.8±8.7	10.7±6.5	−1.1	0.248
HbA1c (%)	5.7±0.8	5.7±0.6	0.0	0.006*

Notes: Values are presented as mean ± SD. Wilcoxon signed-rank test was use for analysis. *P<0.05.
Abbreviations: ALT, alanine transaminase; AST, aspartate transaminase alanin



European Reference Networks



Hartl, D., *et al.* . *J Transl Med* **19**, 245 (2021).

CONCLUSIONS

- The ERNs developed a joint strategy for training and education to overcome the gap between the increasing need of awareness on RDs and complex conditions and the lack of a specific training in the medical curricula.

- The ERNs have created a model that could facilitate the set-up of a European Education Programme in the area of rare diseases and complex conditions.

- A strategy and a road map have been designed to protect the future of RDs by developing a curriculum which can contribute to form a new generation of medical professionals prepared to diagnose and assist patients with RDs.

- New challenges must be faced though:
  - The lack of interest for RDs after the pandemics
  - The shortage of medical professionals EU wide
  - The need of mastering new technologies at molecular and in particular at Informatic level (artificial intelligence) which requires a collaboration with other professional profile sometime not so closed to the medical world.