ERNs

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Rare skin disorders

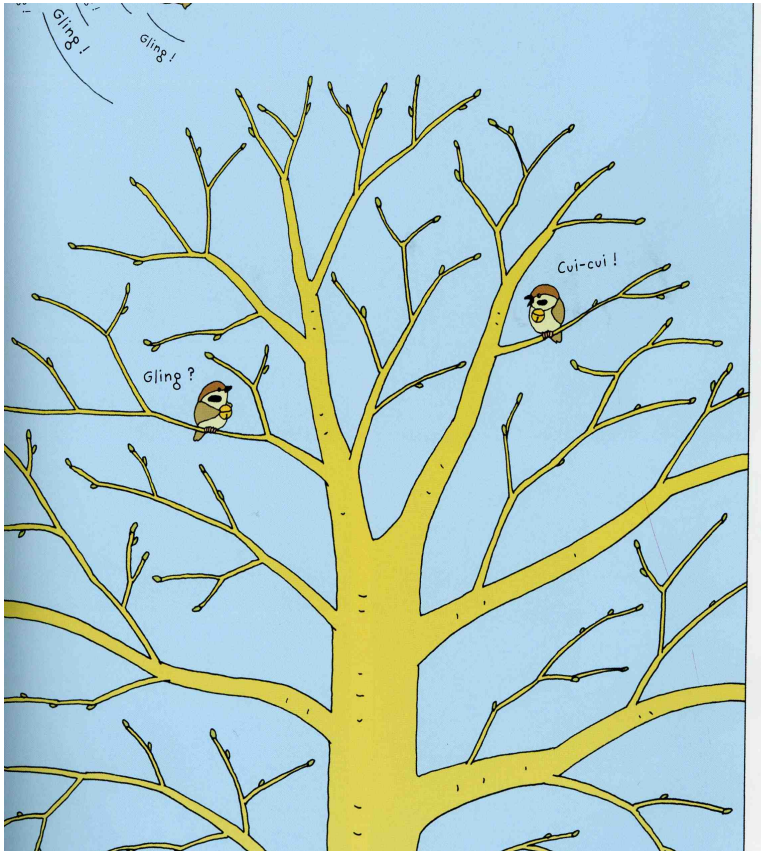
have been identified

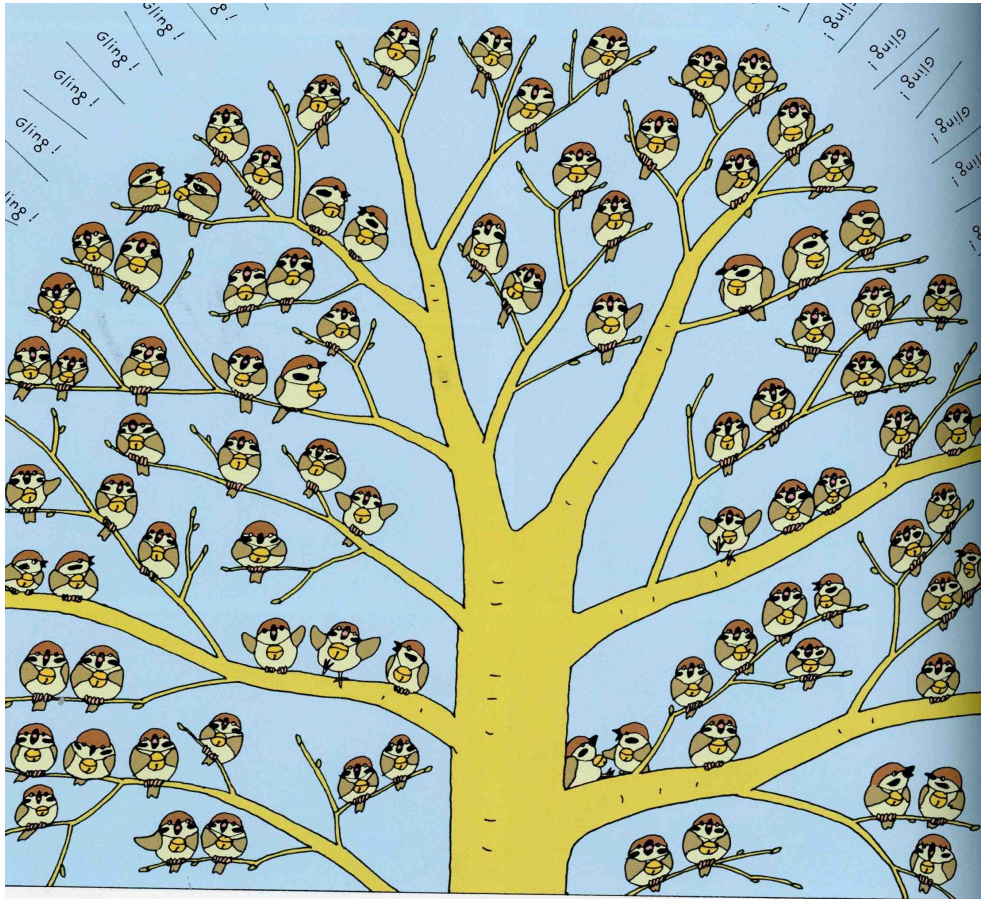
by the *Board of Member States*

on European Reference Network

as a priority area

Maladies rares:
maladies atteignant 1/2 000 personnes en Europe


Gling !
Gling !
Cui-cui !
Gling ?


Gling !
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What is an ERN?

European Reference Networks

An initiative of

European Commission

«Imagine …

- *if the best specialists*
- *from across Europe*
- *could join their efforts*
- *to tackle complex or rare medical conditions*
- *that require highly specialised healthcare and a concentration of knowledge and resources»*


ERN - National - international collaboration
High Level Patient Management
Teaching and Training
IMPROVE THE QUALITY AND SAFETY AND ACCESS TO HIGHLY SPECIALISED HEALTHCARE
Communication e-Health Tools
Research Registries
Patient Empowerment ePAG
Outcome measures and quality control

Key role of Patient Groups

- To enable the full engagement of patient groups through democratic representation, support and quality contribution,

- Eurordis has organized **Policy Action Groups** based on the “ERN disease grouping”

ERN tentative timeline & milestones

European Commission

March – June 2016

Call for Networks

July-October 2016

Assessment proposals

October -November 2016

Approval ERN by Board MS

November 2016 – March 2017

Grant evaluation

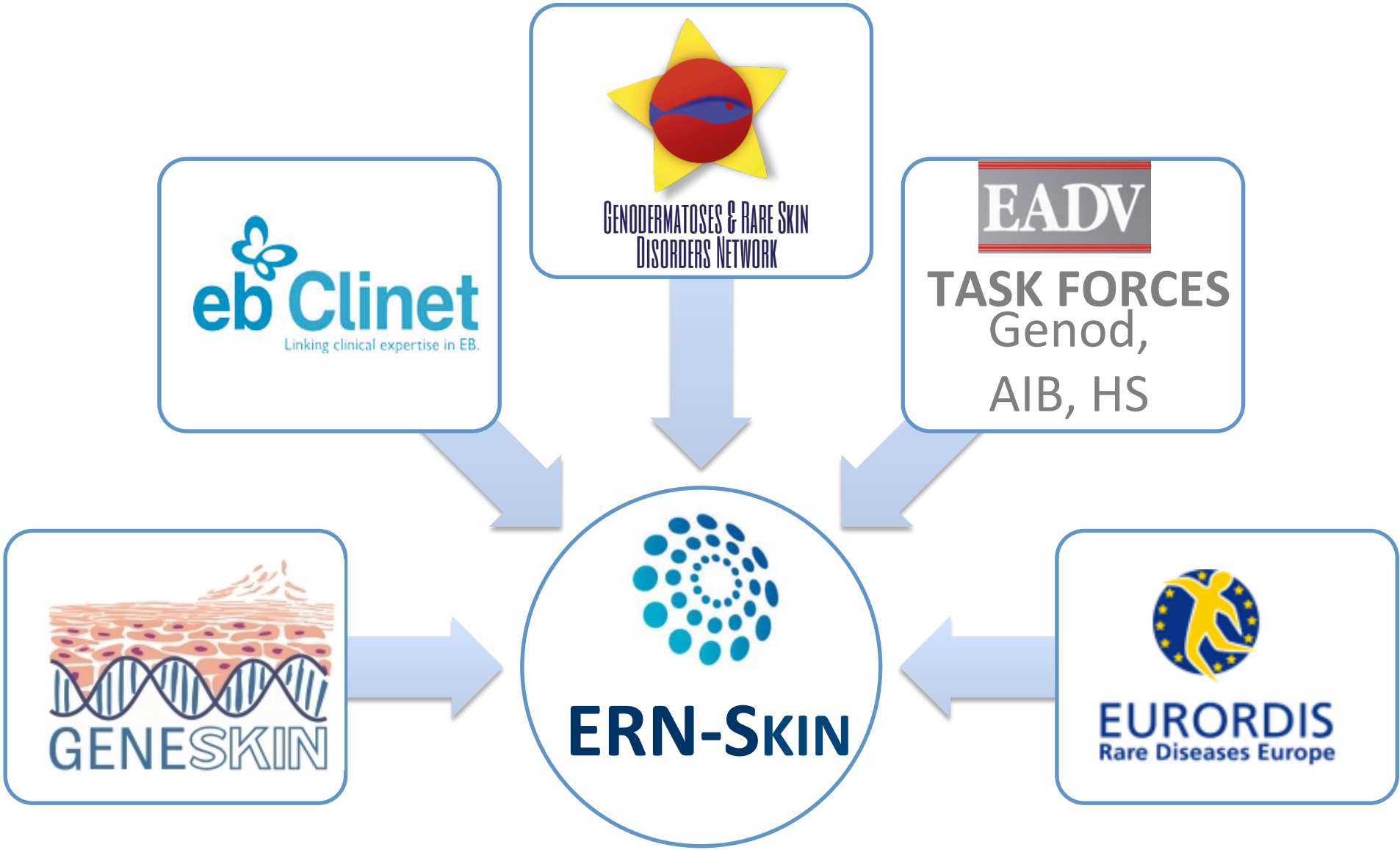
March 2017

3rd ERN Conference & Kickoff meeting ERNs

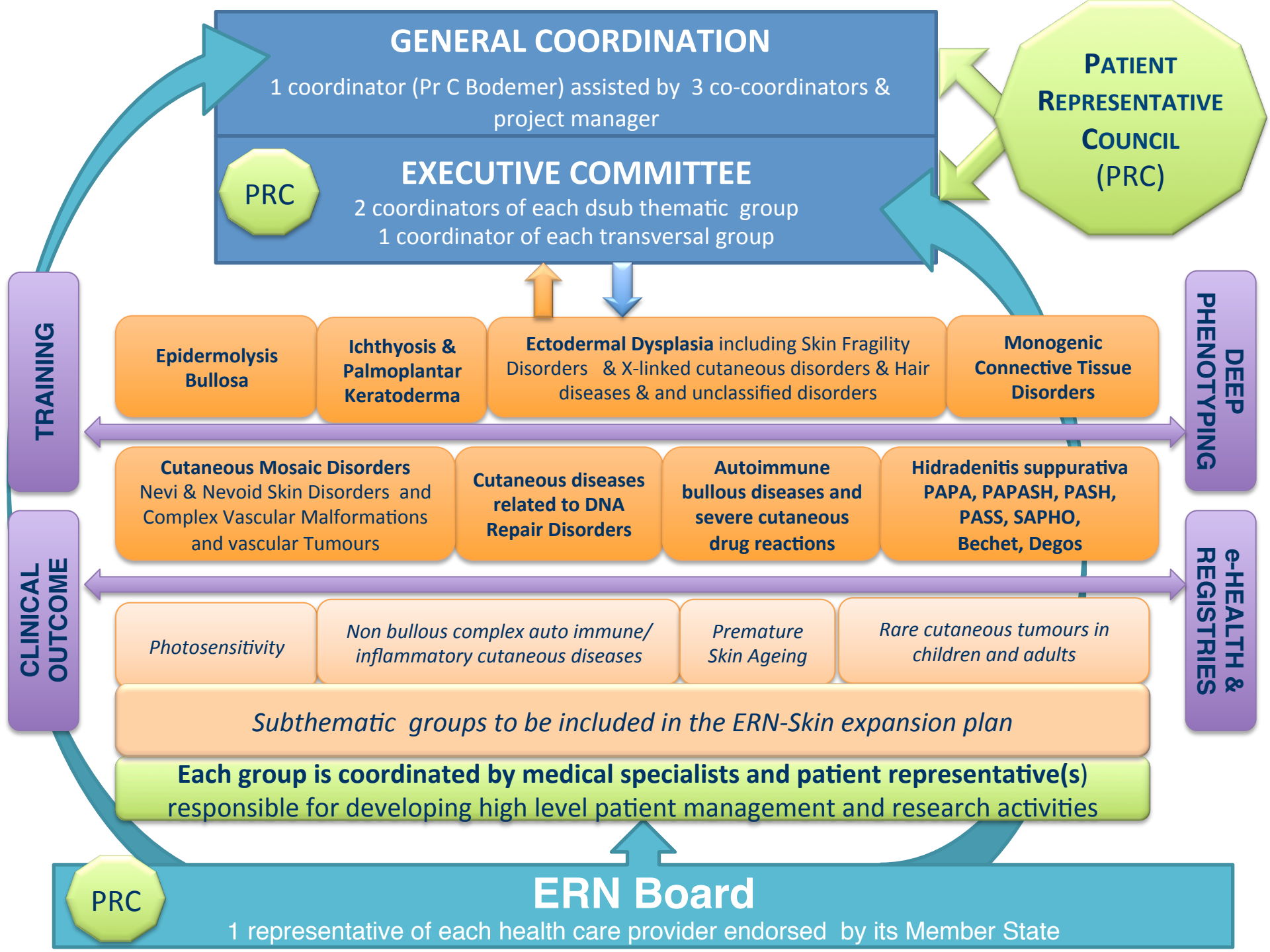
Health and Consumers

European Reference Networks

24 ERNs



Rare and Undiagnosed Skin Disorders


GENERAL COORDINATION
1 coordinator (Pr C Bodemer) assisted by 3 co-coordinators & project manager
PRC
EXECUTIVE COMMITTEE
2 coordinators of each dsub thematic group
1 coordinator of each transversal group
PATIENT REPRESENTATIVE COUNCIL (PRC)
TRAINING
CLINICAL OUTCOME
DEEP PHENOTYPING
e-HEALTH & REGISTRIES
Epidermolysis Bullosa
Ichthyosis & Palmoplantar Keratoderma
Ectodermal Dysplasia including Skin Fragility Disorders & X-linked cutaneous disorders & Hair diseases & and unclassified disorders
Monogenic Connective Tissue Disorders
Cutaneous Mosaic Disorders
Nevi & Nevoid Skin Disorders and Complex Vascular Malformations and vascular Tumours
Cutaneous diseases related to DNA Repair Disorders
Autoimmune bullous diseases and severe cutaneous drug reactions
Hidradenitis suppurativa
PAPA, PAPASH, PASH, PASS, SAPHO, Bechet, Degos
Photosensitivity
Non bullous complex auto immune/ inflammatory cutaneous diseases
Premature Skin Ageing
Rare cutaneous tumours in children and adults
Subthematic groups to be included in the ERN-Skin expansion plan
Each group is coordinated by medical specialists and patient representative(s) responsible for developing high level patient management and research activities
PRC
ERN Board
1 representative of each health care provider endorsed by its Member State

12 SUB THEMATIC GROUPS

Epidermolysis Bullosa	**Ichthyosis & Palmoplantar Keratoderma**	**Ectodermal Dysplasia** including Skin Fragility Disorders & X-linked cutaneous disorders	**Monogenic Connective Tissue Disorders**
Cutaneous Mosaic Disorders Nevi & Nevoid Skin Disorders & Complex Vascular Malformations & Vascular Tumours	**Cutaneous diseases related to DNA Repair Disorders**	**Autoimmune bullous diseases and severe cutaneous drug reactions**	**Hidradenitis suppurativa PAPA, PAPASH, PASH, PASS, SAPHO, Behcet, Degos**
Photosensitivity	*Non bullous complex autoimmune/ inflammatory cutaneous diseases*	*Premature Skin Ageing*	*Rare cutaneous proliferation in children and adults*

Subthematic groups to be included in the ERN-Skin expansion plan

ERN-SKIN

Sub thematic groups with undiagnosed disorders

The Rational

-The complex pathophysiologic mechanisms of the different sub thematic groups often match each other:
the better understanding of some skin
diseases will be a key to have a better understanding of the others, providing a better knowledge on the skin biology

-Each sub thematic group needs a multidisciplinary approach objective to develop a collaborative approach with other ERNs

-A lot of unclassified patients

ERN-SKIN

Sub thematic groups with undiagnosed disorders

The Rational

-Misdiagnosis (poor skin knowledge among HCP)
-Ignorance regarding the management of skin symptoms inflammation, sbarrier alteration, malnutrition, sepsis, pain
-Lack of training of paramedical teams
-Poor recognition of the skin alteration as a handicap
-Poor social integration of patients
-Life-threatening diseases, skin carcinogenesis due to inflammation (chronic wounds…), immunosuppression or genetic predisposition, secondary effect of treatments
-The cost of treatments

Maladies rares

« Les sans diagnostics »

Ont un « village »

Importance d'une description clinique précise
« deep Phenotyping »
Importance des centres experts
même langage (codage: Dg, symptômes)
échanges (ERN-IT plateforme)

Les classifications évoluent intégrant maladies/syndromes anciennement décrits et maladies/syndromes nouveaux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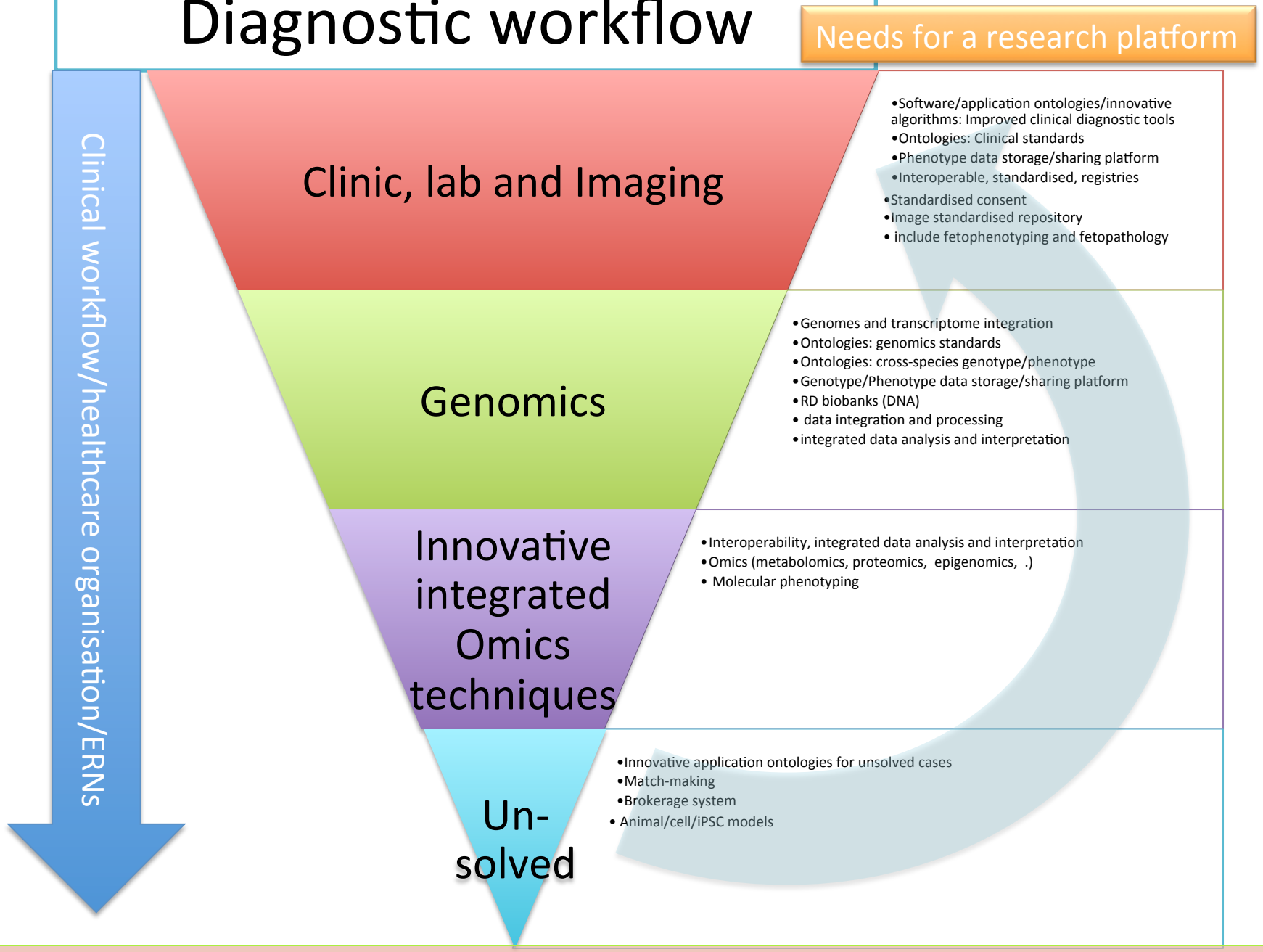
démembrement clinique/physiopathologique/génétique

Intrication étroite de la recherche clinique et de la recherche fondamentale: translationnel au bénéfice du patient

Diagnostic workflow



Interoperability between clinical repositories and research platforms/data sharing standards

Merci