Transforming rare diseases research through transnational funding



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A Rare International Dialogue RDI-CORD Conference 10 – 12 of May, Toronto, Canada

IRDiRC Goals 2017–2027

Released 9 August 2017

VISION: Enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention

GOAL 1: All patients coming to medical attention with a suspected rare disease will be diagnosed within one year if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline.

GOAL 2: 1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options.

GOAL 3: Methodologies will be developed to assess the impact of diagnoses and therapies on rare disease patients.



IRDiRC Consortium Assembly Representation



IRDiRC membership evolution from May 2011 to March 2018 (Cumulative growth)



- 31 funders, including CIHR
- o 11 companies
- 13 patient advocates organizations including CORD

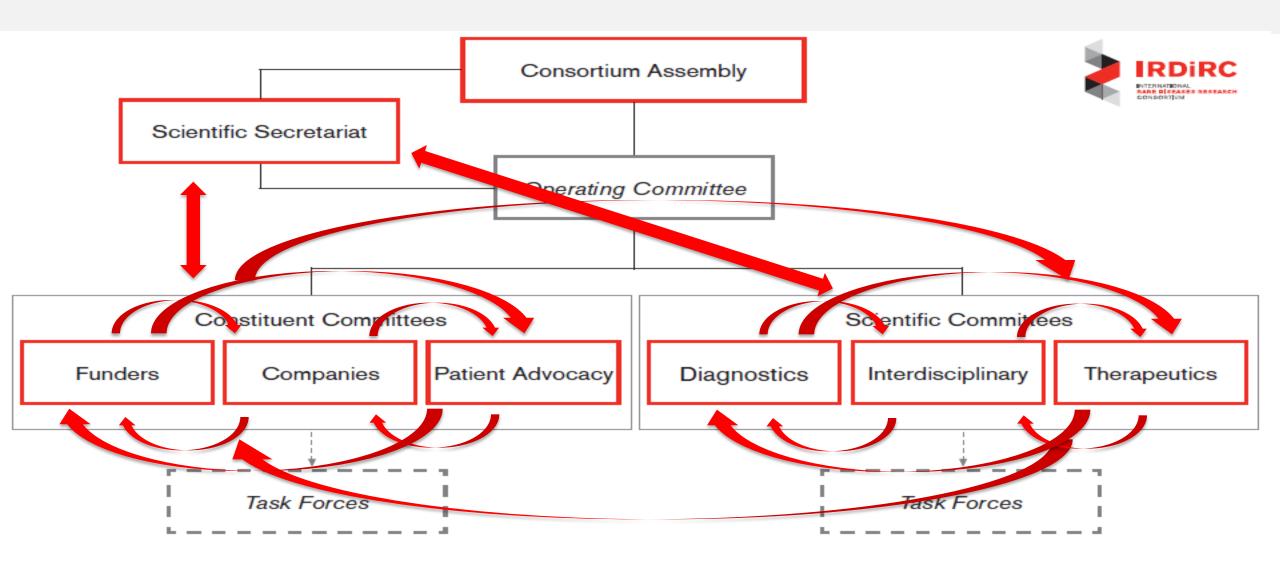


IRDiRC Committees *Mission*

- ► Identify roadblocks/priorities
- ▶ Implement Task Forces and activities to address priorities/gaps
- Establish and promulgate best practices, operating procedures, quality standards, roadmap to address priorities
- Inform other Committees of scientific and programmatic states, needs, opportunities, emerging issues



IRDiRC Structure



More information on IRDiRC

- ► Chair:
 - ♦ Lucia Monaco
- ► Vice Chair:
 - David Pearce
- Scientific Secretariat:
 - Project managers: Carla D'Angelo & Galliano Zanello
 - ♥ Coordinator: Daria Julkowska
- ► Contact:
 - \$\scientificsupport@irdirc.org



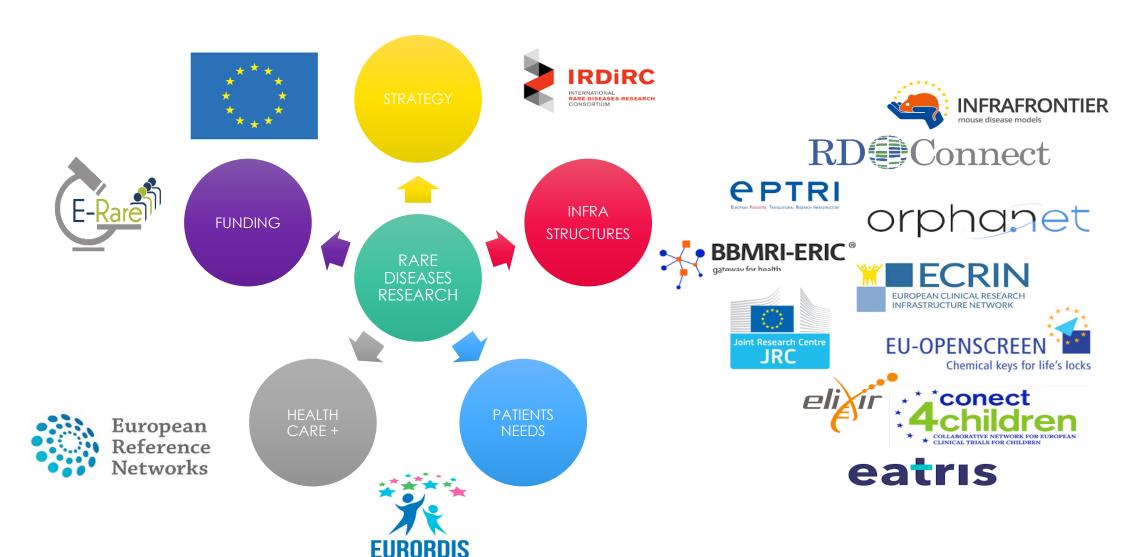


What about Europe?





Rare Diseases Landscape in Europe







Participation of Canada



- **CIHR** and FRQS joined E-Rare ERA-Net in 2013
- Both funding organisation participated in 6 joint transnational calls (2013 - 2018)
- **26** projects with Canadian teams were financed for the total budget of 6.75 M€ (4.6 M CAD)
- 32 Canadian teams were financed
- Muscular Dystrophy Canada financed French research team in 2014 (budget 220 K€)





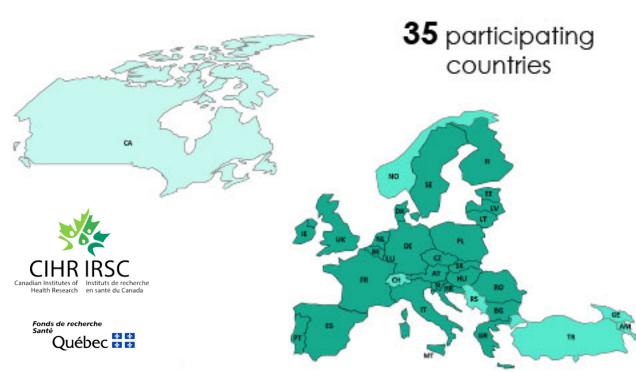


Main facts about the EJP RD

Jan 2019 Dec 2023

Total budget (min. submitted): **101 M** \in (\rightarrow expected > 110 M \in)

Union contribution: 55 M€ (70% reimbursement rate)



27 EU MS (AT, BE, BG, CZ, DE, DK, ES, EE, FI, FR, GR, HU, HR, IE, IT, NL, LT, LV, LU, MT, PL, PT, RO, SE, SK, SI, UK), 7 associated (AM, CH, GE, IL, NO, RS, TK) and CA

88 beneficiaries

- 31 research funding bodies/ministries
- 12 research institutes
- 22 universities/hospital universities
- 🐹 11 hospitals
- 5 EU infrastructures (BBMRI, EATRIS, ECRIN, ELIXIR, INFRAFRONTIER) + EORTC
- 🐹 EURORDIS & ePAGs
- 5 charities/foundations (FTELE, AFM, FFRD, FGB, BSF)
- + 50 Linked Third Parties







COORDINATION & TRANSVERSAL ACTIVITIES

INTEGRATIVE RESEARCH STRATEGY

SUSTAINABILITY

ETHICAL & REGULATORY

COMMUNICATION

1

FUNDING

COORDINATED
ACCESS TO
DATA &
SERVICES

2

3

CAPACITY
BUIDLING &
EMPOWERMENT

ACCELERATING
TRANSLATION
OF RESEARCH &
THERAPY
DEVELOPMENT

4







Pillar 1: Collaborative research funding



Pillar 1: Activities

WP6: Joint Transnational Calls for collaborative research projects



Open to research teams from countries with funders involved – min of 4 teams from 4 countries. Canadian teams are eligible according to the CIHR & FRQS rules. Canadian Pateint Advocacy Organisation (PAO) can receive funding

WP7: Networking to share knowledge on rare diseases



Small support schemes for networking (workshops/events/share of knowledge) – 30 K€ max – open all year long – open to all countries involved in EJP RD including Canada

WP8: Rare disease research challenges



Public-private (small scale) partnerships – challenges set by industry and validated by EJP RD – short term (max. 18 months) projects - open to all countries involved in EJP RD including Canada

WP9: Monitoring of funded projects



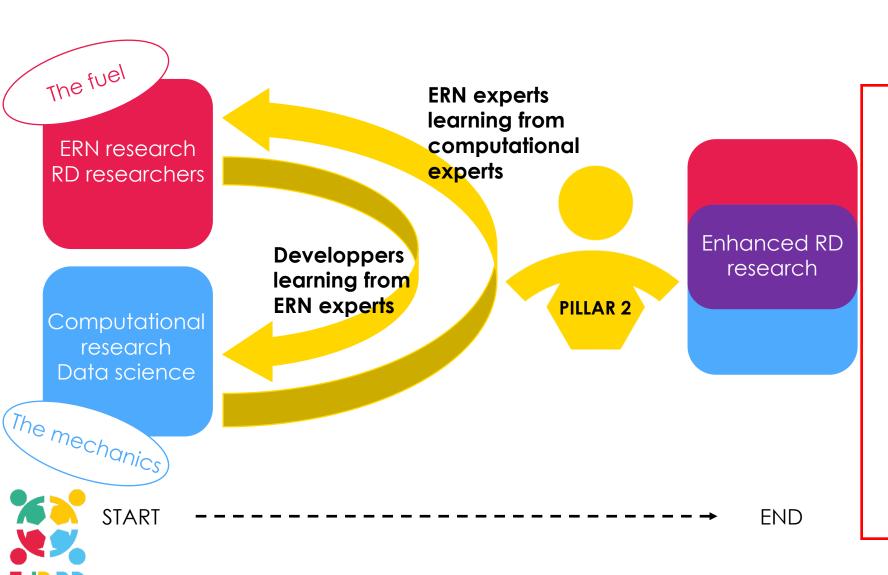
Monitoring of all projects funded through EJP RD and previous E-Rare projects



Pillar 2: Innovative coordinated access to data and services for transformative rare diseases research



Pillar 2 target: FAIR-based virtual platform



A powerful substrate for translational research:

- Centralized services for collections (resource-level)
- Sample, biobanks, registries, infrastructures and tools catalogue
- Analysis platform for omics data
- Curated rare disease-centered information and data
- Federated services for data elements (record-level)
- FAIR 'at source'
- Data, patients, and samples linked and discoverable
- Consents and data use conditions also represented



Pillar 2: Activities



WP10: User-driven strategic planning and transversal activities for Pillar 2 data ecosystem

Annual strategic meetings with users (ERNs) & dev<mark>elopers to define the priorities – coordination of outputs & needs – technical GDPR implementation – quality, sustainability and scaling up</mark>



WP11: Common virtual platform for discoverable data and resources for RD research

Metadata & ontological models – FAIR compliance – data deposition & access to data infras – online tools

WP12: Enabling sustainable FAIRness and Federation at the record for RD data, patients and samples

Alignement of core interoperability standards – software for FAIR ecosystem – FAIRification support

WP13: Enabling multidisciplinary, holistic approaches for rare diseases diagnostics and therapeutics



System biology approaches for RD – biological pathways – variants to function – environmental toxicology – treatment drugs - proof of principle studies





Pillar 3: Capacity building and empowerment



Pillar 3: Activities

WP14: Training on data management & quality

Orphanet nomenclature – standards & quality of genetics/genomics data in clinical practice – strategies to foster undiagnosed diseases – biobanks sample data management – rare diseases registries & FAIRification at source – European Rare Diseases Registry Infrastructure

WP15: Capacity building & training of patients and researchers in rare diseases research and processes

Expert Patients and Researchers EURORDIS Summer school – scientific innovation and translation research aspects in RDs for patient advocates – leadership & communication skills for patient advocates and representatives – education material and activities for paediatric patients

WP16: Online academic education course

Based on assessed needs of the RD community – in collaboration with universities – 10 to 12 modules with accreditation – e-learning format open to all – Future Learn platform

WP17: ERN RD training & support programmes

Based on four groups (Neuro, Neoplasm & malformation, Organs, Systemic) – preferences, needs and resources of ERNs – tailored for and performed by ERNs



WP18: Development and adaptation of training activities

Evaluation of developing needs according to progress of Pillars 2 & 4 – specific needs of EU 13 countries – emerging needs of ERNs

Pillar 4: Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations



Pillar 4: Activities

WP19: Facilitating partnerships and accelerating translation for higher patient impact

Innovation management toolbox – assessment and real time mentoring of translational projects – support in exploitation and follow-on funding – partnering support – roadmap for European investment platform for RD

WP20: Accelerating the validation, use and development of innovative methodologies tailored for clinical trials in RDs

Key Task Force group - Support in design and planning of RD clinical studies with ECRIN – demonstration projects on existing statistical methodologies to improve RD clinical trials – innovative methodologies to improve RD clinical trials in limited populations





Conclusions

- Strong partnership with Canadian funding organisations opened new opportunities for rare diseases researchers:
 - Collaborative projects on rare diseases
 - Access for CA researchers and PAO to funding, tools and support provided within the EJP RD
 - Share of knowledge and perspective beyond Europe
- More direct involvement of Canadian research performing organisation would be profitable to further enhance common actions and bring international dimension to e.g. RD clinical research networks

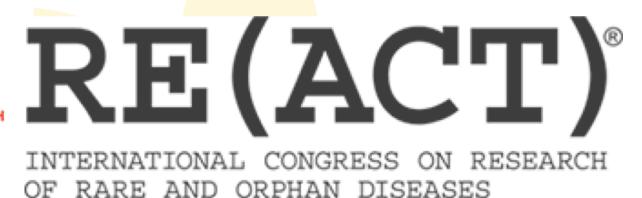




Joint Congress



INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM



****** WHEN: **March 2020**

WHERE: Berlin, Germany

PROGRAMME: dedicated workshops, scientific sessions, policy, patients

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