Breakout session - WG8 Rare Diseases

1+MG / B1MG Stakeholder Forum

Chair: Katrin Õunap

Co-chair: Ruben Kok

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Rare Diseases: Outcomes expected from the 1+MG initiative

- Improved knowledge of the genetic make-up of RDs (disease-genes discovery)
- Anticipation of diagnosis and solving the unsolved
- Better understanding of the phenotypic variability and longitudinal course of RD;
- Support clinicians to make decisions on how to adapt treatment and care pathways.

Addressing the needs of RD research

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17 MSs & EJP-RD have designated 31 experts in WG8

Bulgaria	Rumen Stefanov
Czechia	Milan Macek
Denmark	Irene Kibæk Nielsen
Estonia	Katrin Õunap, Sander Pajusalu
Finland	Kuismin Outi
Germany	Olaf Rieß, Dagmar Wieczorek
Greece	Periklis Makrythanasis
Italy	Bruno Dallapiccola, Marco Tartaglia
Latvia	Viktorija Ķēniņa, Baiba Lāce
Lithuania	Lina Jankauskaitė, Birutė Tumienė, Edita Baltruškevičienė
Luxembourg	Barbara Klink
Malta	Joanna Vella, Alex Felice
Netherlands	Lisenka Vissers, Gijs Santen
Portugal	Laura Vilarinho
Slovenia	Luca Lovrečić, Ana Torkar, Nataša Debeljak
Spain	Pablo Lapunzina
Sweden	Anna Lindstrand, Hans Ehrencrona
EJP-RD	Alain Verloes, Héléne Dollfus

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Scope of WG8

as in the 2018 1+MG declaration

(1) To describe the value of European level data sharing.

(2) To describe how shared data can be used within the RD community

(3) To consider the needs of both end-users/patients, research, healthcare and industry, taking into account aspects related to disease-gene discovery, mechanism study, diagnostics, therapy, prevention and knowledge building.

(4) To identify ongoing national/European pilot projects.

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Scope of WG8

as in the 2018 1+MG declaration

(5) To make available an inventory of accessible RD data (building on data generated through the mapping exercise): what is currently in place, what is on-going and what is needed.

(6) To provide suggestions on possible synergies with other initiatives.

(7) To provide support to other WGs, in order to reach the goals of the 1+MG roadmap.

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Activity

(1-3) Value/modalities of data sharing/needs in healthcare & research

Key aspects: diagnosis & counseling/molecular basis of disease, gene-specific clinical variability & heterogeneity/allelic disorders & gen-phen correlations, natural history of diseases, call for patients (clinical trials), functional annotation of genomic variants.

Interaction with WG2, WG5 and WG6:

(i) Signed consents, secondary use of clinical/genetic data, transition between healthcare and research (WG2/WP2);

(ii) Use of the infrastructure/access to data (WG5/WP4);

(iii) WGS vs WES in routine diagnostics/research, cost-effectiveness in healthcare (WG6).

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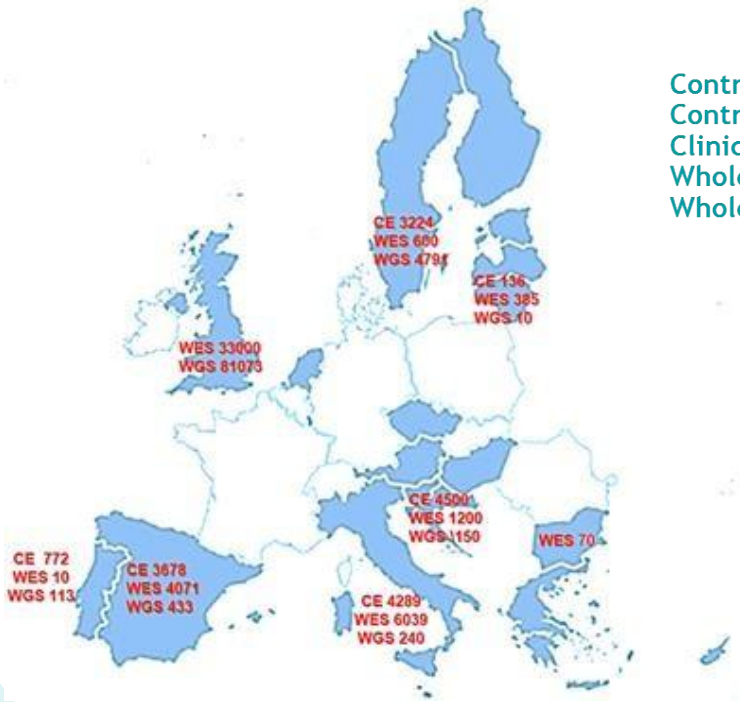
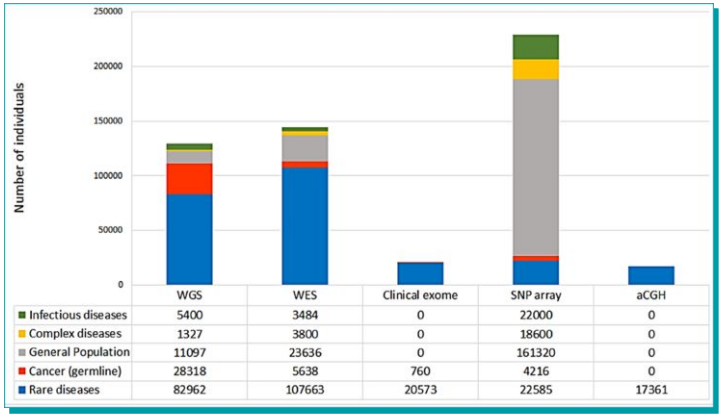
Activity

(5) Inventory of accessible RD data.



2019 Census

Contributing Countries	8/13
Contributing Centers	40
Clinical Exomes (CE)	16,669 (70x - 150x)
Whole Exome Sequencing (WES)	45,305 (40x - 150x)
Whole Genome Sequencing (WGS)	86,809 (8x - 30x)



	WGS	WES	Clinical exome	SNP array	aCGH
Infectious diseases	5400	3484	0	22000	0
Complex diseases	1327	3800	0	18600	0
General Population	11097	23636	0	161320	0
Cancer (germline)	28318	5638	760	4216	0
Rare diseases	82962	107663	20573	22585	17361

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Activity

(7) To support the other WGs to reach the goals of the 1+MG roadmap.

15/10/2020 – Healthcare Use Case Workshop (virtual meeting)

07/12/2020 - Synthetic data workshop (virtual meeting)

29/04/2021 - Research Use Case Workshop (virtual meeting)

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Activity

(4,6) To promote synergies with other ongoing projects.

2019

28/06 - Amsterdam, F2F meeting with Daria Julkowska, EJP-RDCoordinator, for possible collaborations between EJP-RD and 1MG WP8.

26/07, 28/08, 30/09 - Virtual meetings, CSA Genomics & Personalised Medicine Proposal (SC1-HCC-06-2020).

2020

21/10 - First 1+MG / B1MG Stakeholder Forum (virtual meeting).

2021

14/03 - Rare Disease use case alignment across ELIXIR - B1MG -1+MG - EJP-RD workshop (virtual meeting)

17/11 - Second 1+MG / B1MG Stakeholder Forum - Breakout 1, Rare Diseases (virtual meeting)

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Activity (other activities/meetings/workshops)

2019

5-6/06 - Brussels, Meeting of WG8 experts and the Representatives of the Signatories of the Declaration.

17-18/09 - Gdansk, EJP-RD General Assembly Meeting: presentation of the 1+MG project.

11/11 & 25/11 - 1+MG virtual meeting of WG coordinators.

10/12 - Brussels, 1+MG WG leaders + core group.

June-December - Delivery/circulation of a WG8 document addressing the main planned deliverables (available at the EC CIRCABC site).

2020

20/01 - 1+MG virtual meeting of WG coordinators.

04/02 - Brussels, 7th Meeting of the Representatives of the Signatories of the 1+MG Declaration.

24/04, 29/05, 26/06, 31/07, 28/08, 25/09, 30/10, 27/11 - Virtual meetings of the 1+MG coordinating team and WGs' leaders.

10/05 - The 1+MG roadmap circulated among the designated WG8 experts.

26/11 - Signatory MS virtual meeting.

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Activity (other activities/meetings/workshops)

2021

29/01, 26/02, 26/03, 30/04, 25/06, 24/09, 29/10 - Virtual meetings of the 1+MG coordinating
team and WGs' leaders.
18/03 - 1+MG Signatory MS Group virtual meeting.
23/03 - B1MG WP5 Country Exchange Visit - United Kingdom (virtual meeting).
21/04, 06/10, 30/11 - WG8 experts' meetings (virtual meetings)
18/11 - Virtual B1MG General Assembly Meeting

2022

28/01, 25/02 - Virtual meetings of 1+MG coordinating team and WGs' leaders.
21/02, 11/03 - WG8 experts' meetings (virtual meetings)
08/03 - B1MG third EC review virtual meeting.

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Current status

- PoC (architecture of federated infrastructure), standards, approaching ELSI

Next steps

- Access to the federated data, generation of data

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Open to discussion

Gaps and pitfalls	Recommendations
Expected timeline to have **access to the federated Infrastructure** and be able to perform individual queries	In RDs, research and healthcare are often done in conjunction. Consider a single access path for RD diagnosis and novel pathogenic gene identification.
Existence of **large collections of WES data** from "unique" patients/diseases	Define WES data standards also (not only WGS) It should be considered how to include them in the 1+MG IT
Connection of the 1+MG IT and datasets to other International Initiatives	Have bi-directional data discovery mechanisms connection to other resources or IT networks (e.g. MatchMaker Exchange nodes outside Europe).
RD data is usually longitudinal	Evaluate how to address this from the ELSI and IT.

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Raredisease

Thank you!

You will soon rejoin the main session

Feedback

Breakout session - WG8 Rare Diseases

Chair: Katrin Õunap

Co-chair: Ruben Kok

ELSI & Data Governance: Which challenges to the scale up (within this Use Case) could be addressed by industry?

- Implementation of electronic consent system
- A dynamic consent solution
- Sharing knowledge from multinational studies
- To add the consent form and note of data sharing with industry and stakeholders
- Their collaboration on machine readability of consent and use condition of collected data (different from electronic consent)
- Transparency on how patient data benefits the company. It could for instance help to train algorithms, or help as proof of function to boost sales.
-

ELSI & Data Governance: Which challenges to the scale up (within this Use Case) could be addressed by other stakeholders (please specify stakeholder)?

- Privacy officers in hospitals
- There is a big gap between clinical actors and FAIR principles - understanding, mindset and solutions. Can FAIR ambassadors help - Universities, Funders
- EJPRD; making access conditions machine readable in order to enable easier compliance with ELSI conditions
- Difficult to share with US collaborators as they use google and its not allowed by EU GDPR
- Allow for secondary use of genomic data (data sharing) outside of research studies (EU and government)
- We should know who is addressing what issue for example GA4GH is addressing many of the ethical and legal issues surrounding genomic data.
- National DPBs should come up with suggestions on how data can be used responsibly, and allowing collaboration between industry and health care institutes.

Data & Quality Standards: Which challenges to the scale up (within this Use Case) could be addressed by industry?

- Automated extraction of minimal dataset
  - Have plug-ins from all local hospital systems and facilitate those conversions through tools and software we use
  - Capture data from clinical systems
  - Both ways lab to clinical and vice versa
- Tools to facilitate data entry with HPO, ORDO within EHRs
- Translation of ontologies
- Standards, tools, and requirements developed and adopted by industry
  - https://github.com/ejp-rd-vp

Data & Quality Standards: Which challenges to the scale up (within this Use Case) could be addressed by other stakeholders (please specify stakeholder)?

- HTA and regulators (EMA and EUnetHTA)
  - They are developing their own processes and data spaces and guiding on the type of data
  - EMA through the DARWIN initiative and have provided a lot of guidelines and data
- Usage of tools already implementing standards (eg. EJPRD virtual platform)
- Quality control, accredited lab/centre
- Strong guidelines on data standardisation on EU level, forcing national implementations to follow the EU guidelines.

Technical Infrastructure: Which challenges to the scale up (within this Use Case) could be addressed by industry?

- Support to find “right” fit for integration/ “make whatever we need” adaptable to personal local situation
- On what level are we sharing data with industry
  - Much easier to share data with industry that you aren’t going to return results to an individual person.
  - There are different local regulations if the data is being returned for the benefit of specific individuals
- Industry infrastructures made interoperable with 1+MG Infrastructure
- (Financial/ technical) Support to find “right” fit for integration/ “make whatever we need” adaptable to personal/local situation.

Technical Infrastructure: Which challenges to the scale up (within this Use Case) could be addressed by other stakeholders (please specify stakeholder)?

- EJPRD virtual platform interoperability
- Heterogenous maturity levels among MS in digital data storage and HCP infrastructure
    - Belongs to people who are investigated and the national governing body should decide how to keep that data
- Cloud-based tools and software can be provided locally - no internet access - it requires HPC platforms to engage with providers
- GA4GH toolkits for responsible data sharing (e.g file formats, APIs, standards, recommendations, etc.)
- Critical Path Institute, Data processing/Analysis Platform: https://portal.rdca.c-path.org/

General Comments:

- Workshops (technical hands-on and dissemination/sustainability planning): EJP RD and next phase RD Partnership
- Already mentioned, but many examples have already been piloted, and proven successful (such as EJP-RD and SOLVE-RD)
- Patient initiatives to donate data
- National requirements and resources to build up Germ Line data at clinical level.
- A guidebook containing the relevance resources and tools for researchers just starting or Member States looking to start implementing genomics into healthcare systems.

Thank you!

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