# Webinar | IMI2 - Call 23
# Shortening the path to rare disease diagnosis by using new born genetic screening and digital technologies

29.06.2020

# Agenda

- How to use GoToWebinar – Catherine Brett, IMI
- Introduction – Magda Gunn, IMI
- The Call topic – Nicolas Garnier, Pfizer
- Involvement of SMEs, patient groups, regulators – Magda Gunn, IMI
- Questions & answers

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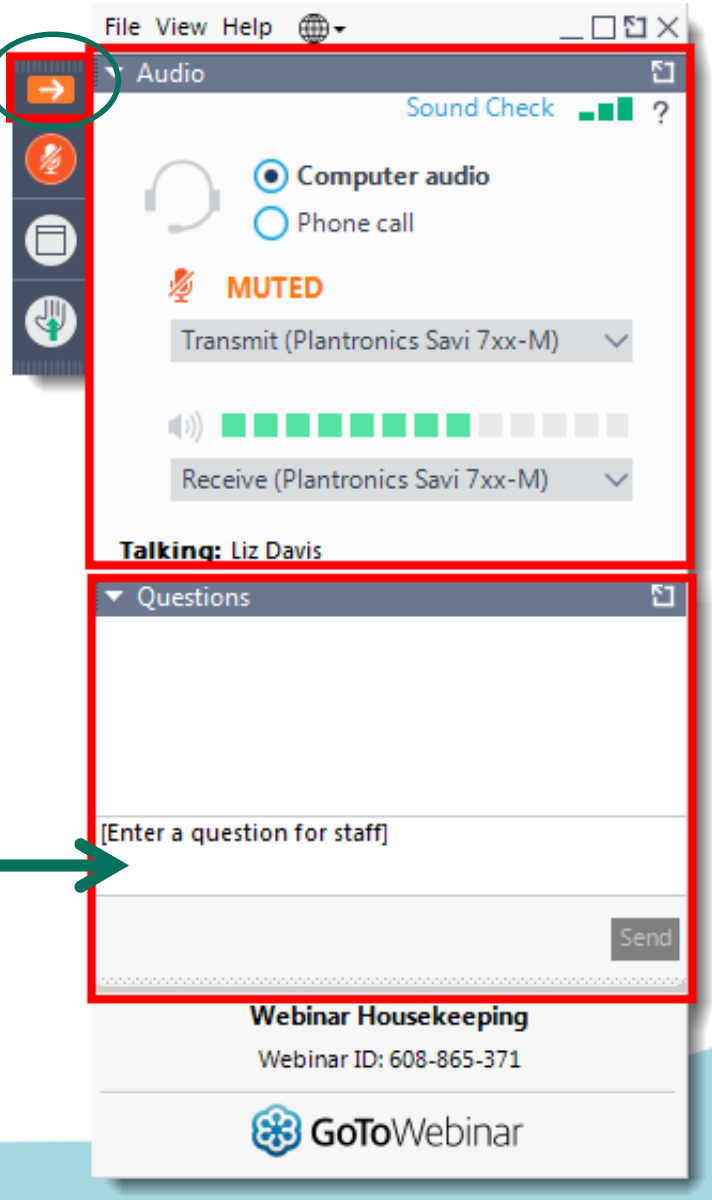


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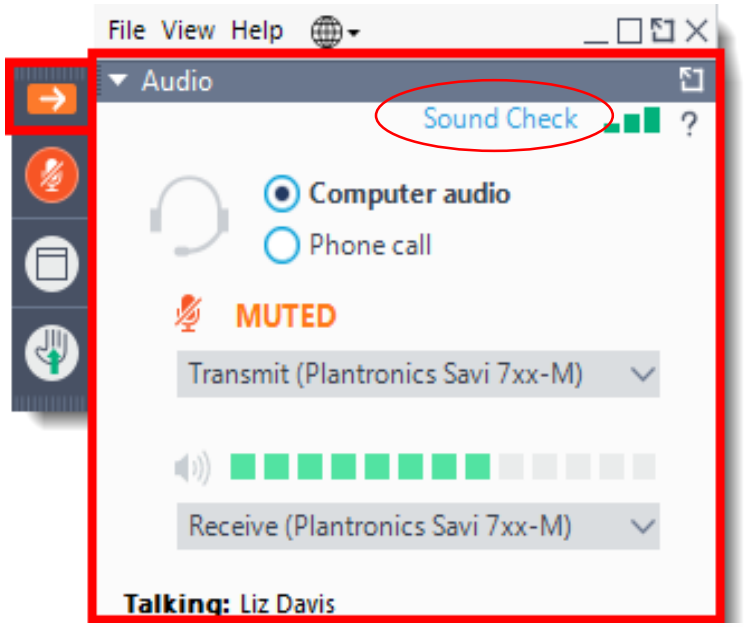


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# Before we start…

- We are recording this webinar and it will be published on the IMI website and / or IMI YouTube channel
- We will also publish the presentation slides and the participant list on the webinar web page
- IMI2 – Call 23 has been launched and all Call documents & details of how to apply can be found on the IMI website

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# Webinar | IMI2 - Call 23

# Shortening the path to rare disease diagnosis by using new born genetic screening and digital technologies

**Magda Gunn**

29 June, 2020

# Today's webinar

**Will cover all aspects of the Call topic**

- Introduction to IMI programme
- Proposed project
  - Objectives, need for public-private collaborative research
  - Key deliverables
  - Structure of the project
  - Expected contribution of the applicants
  - Contribution of industry consortium

**Will not cover rules and procedures**

- A webinar on rules and procedures will take place on
  30 June 2020, 11:00 am – 12:30 pm CEST

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# IMI – Europe's partnership for health

**IMI mission**

IMI facilitates open collaboration in research to advance the development of, and accelerate patient access to, personalised medicines for the health and wellbeing of all, especially in areas of unmet medical need.

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# IMI – Ecosystem for innovative collaborations

- Allow engagement in a cross-sector, multi-disciplinary consortium at the forefront of cutting-edge research
- Provide the necessary scale by combining funding, expertise, knowledge, skills and resources
- Build a collaboration based on trust, creativity and innovative and critical thinking
- Learn from each other - new knowledge, skills, ways of working
- Take part in transformative research that will make a difference in drug development and ultimately patients' lives

IMI is a **neutral platform** where **all involved** in drug development can engage in **open collaboration** on **shared challenges**.

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# IMI partnership 2008-2020

**IMI1:**

- 2008-2013
- €2 bn budget
- 59 projects

**IMI2:**

- 2014-2020
- €3.3 bn budget
- More ambitious, more open, greater scope


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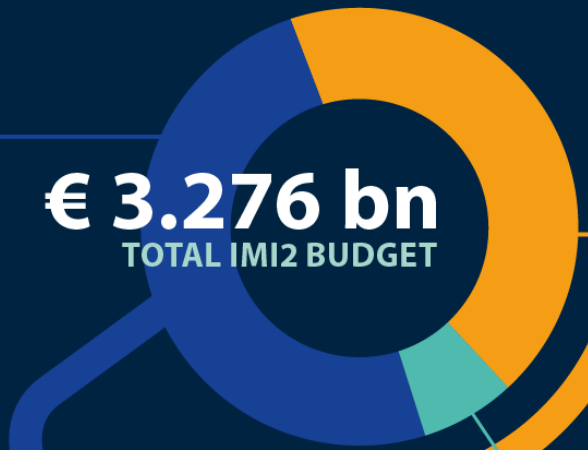
# IMI2 funding
(2014-2020)

IMI FUNDING MODEL

efpia

**IN-KIND PRIVATE CONTRIBUTION**
**€1.425 bn**
EFPIA companies receive no funding

**public contribution**
**€1.638 bn**
funding from Horizon 2020




**OTHER CONTRIBUTIONS**
**€213 MILLION**
(Associated Partners, e.g. charities, non-EFPIA companies)

EU funding goes to

SMES
UNIVERSITIES
PATIENTS, REGULATORS...

EFPIA contribute researchers, laboratories, generation of data, curation of compounds, and cash

**Public and private partners collaborate in IMI2 projects**

Accelerating research and development

Speeding up patient access to innovative treatments

Improving patient outcomes and safety of medicines

# How a topic is generated

Industrial partners align themselves around a real challenge for industry and agree to work together **and commit resources**

New ideas from public sector, universities, SMEs etc. are needed to address the challenge

Scale is a key to success and is provided through IMI funding

Outcomes should be transformative for the industry as well as having a clear “public” value

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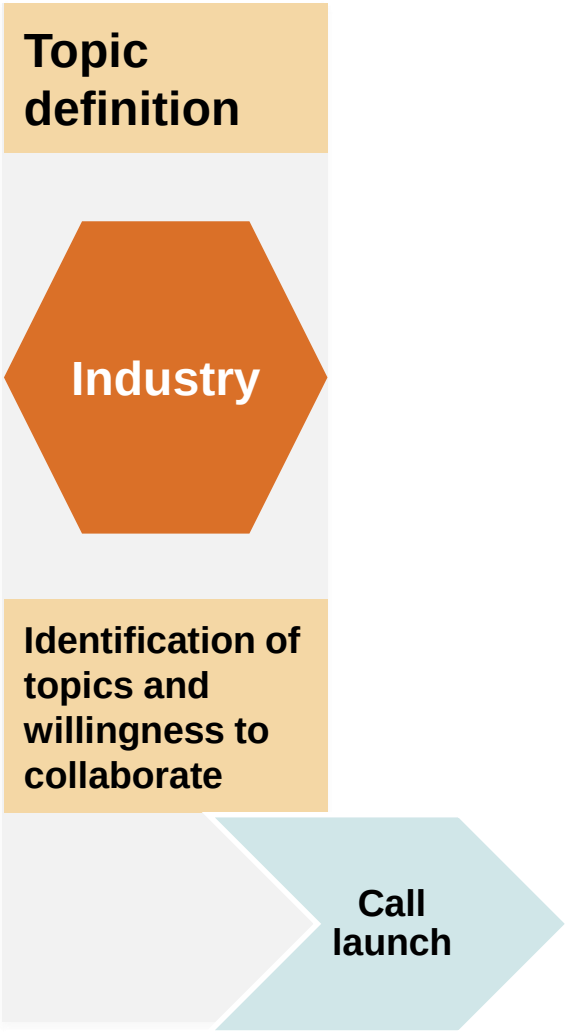
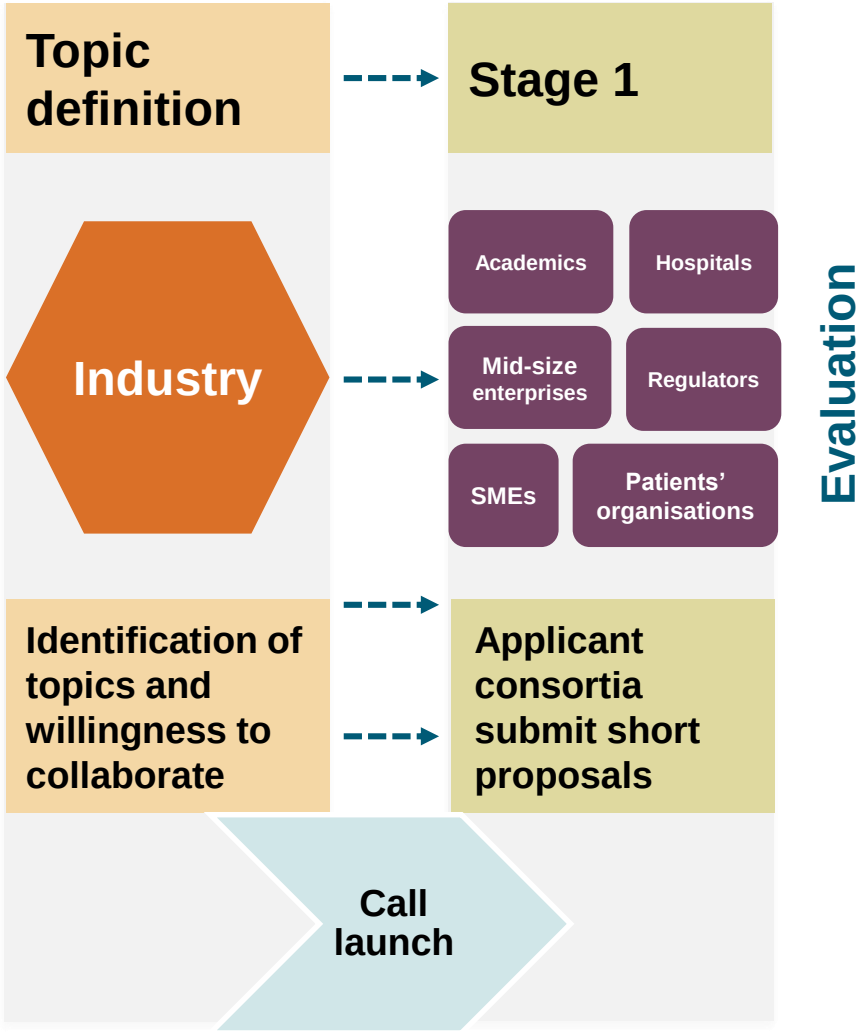
# Typical IMI project life cycle



# Typical IMI project life cycle

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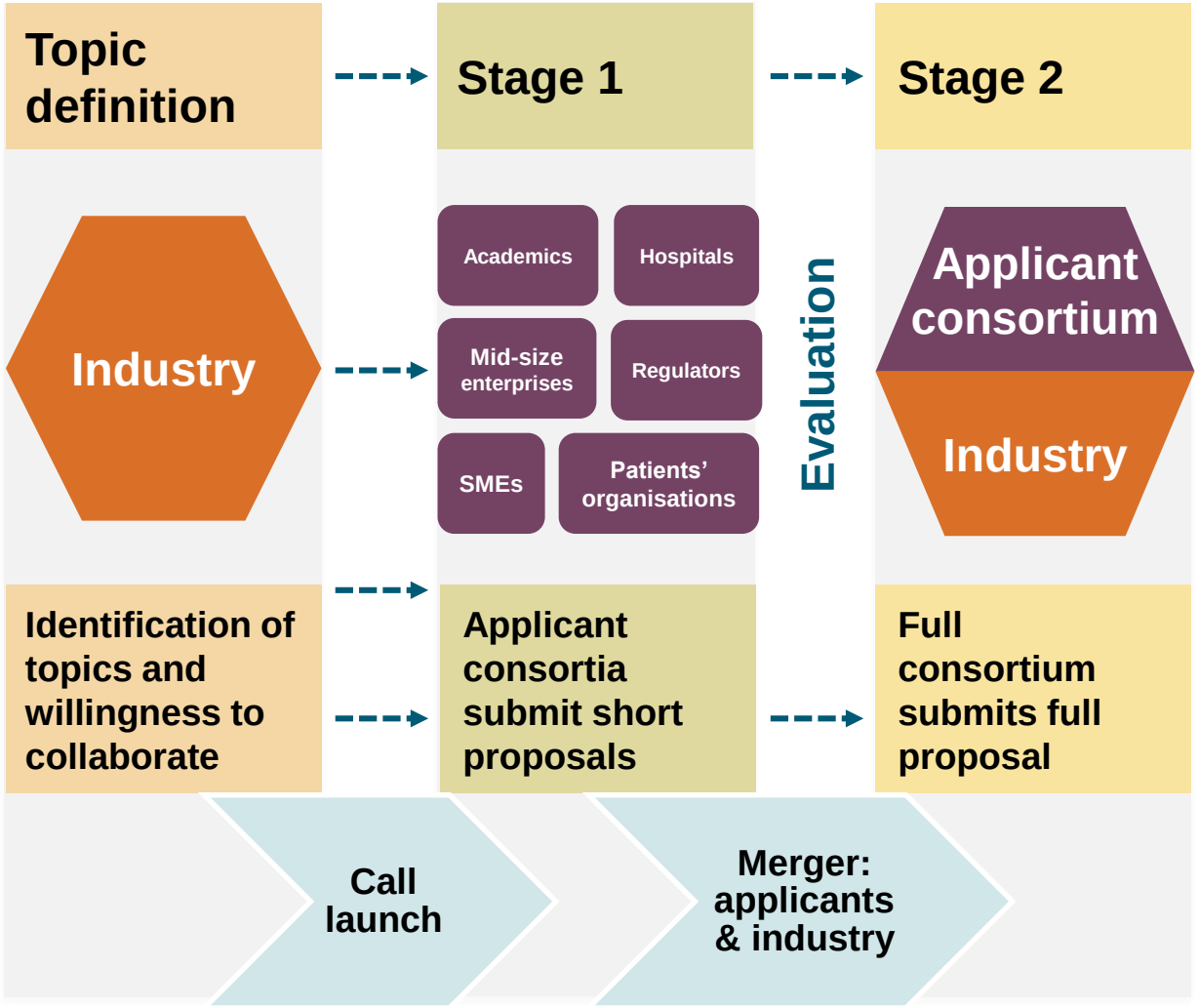
# Typical IMI project life cycle

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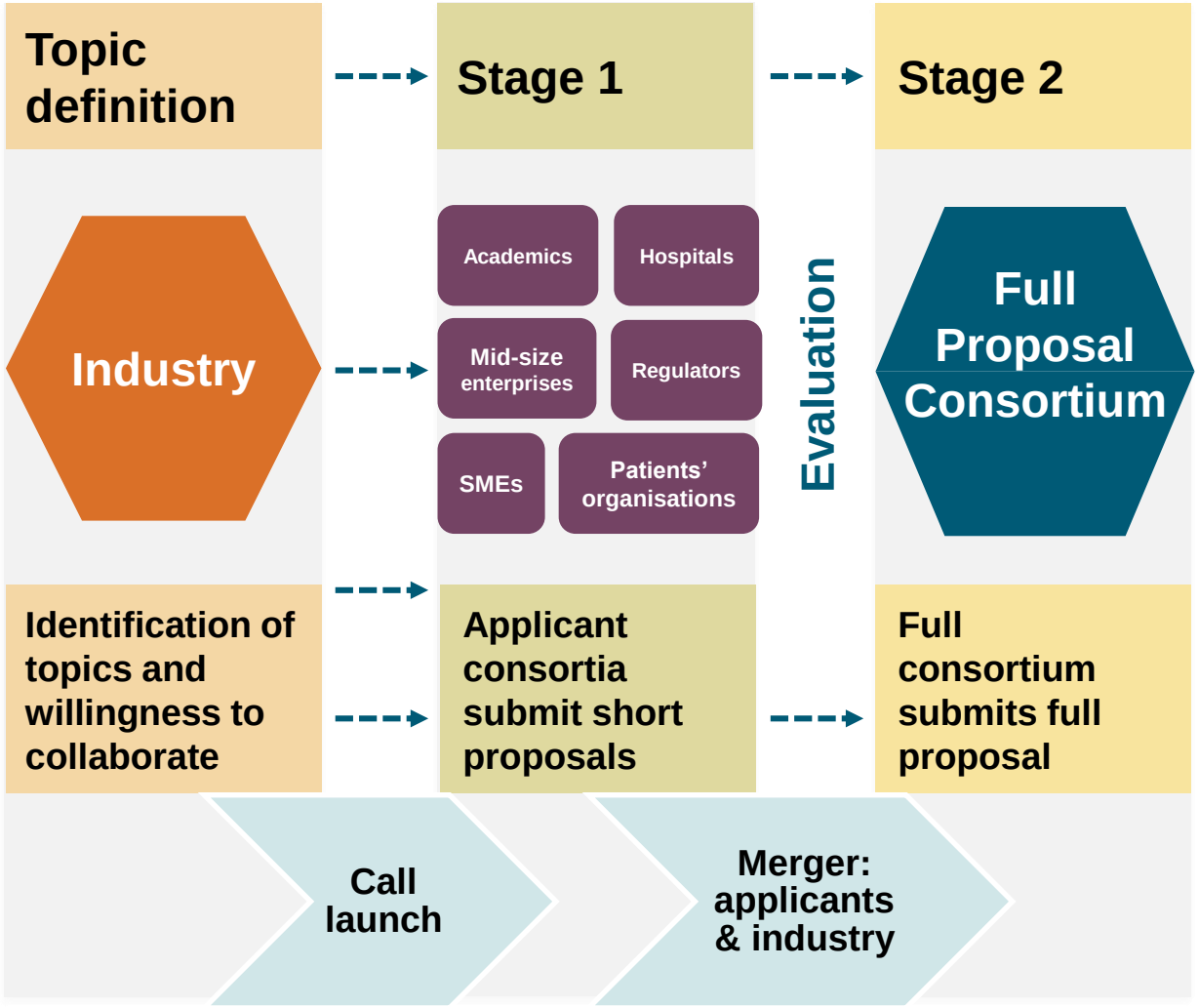


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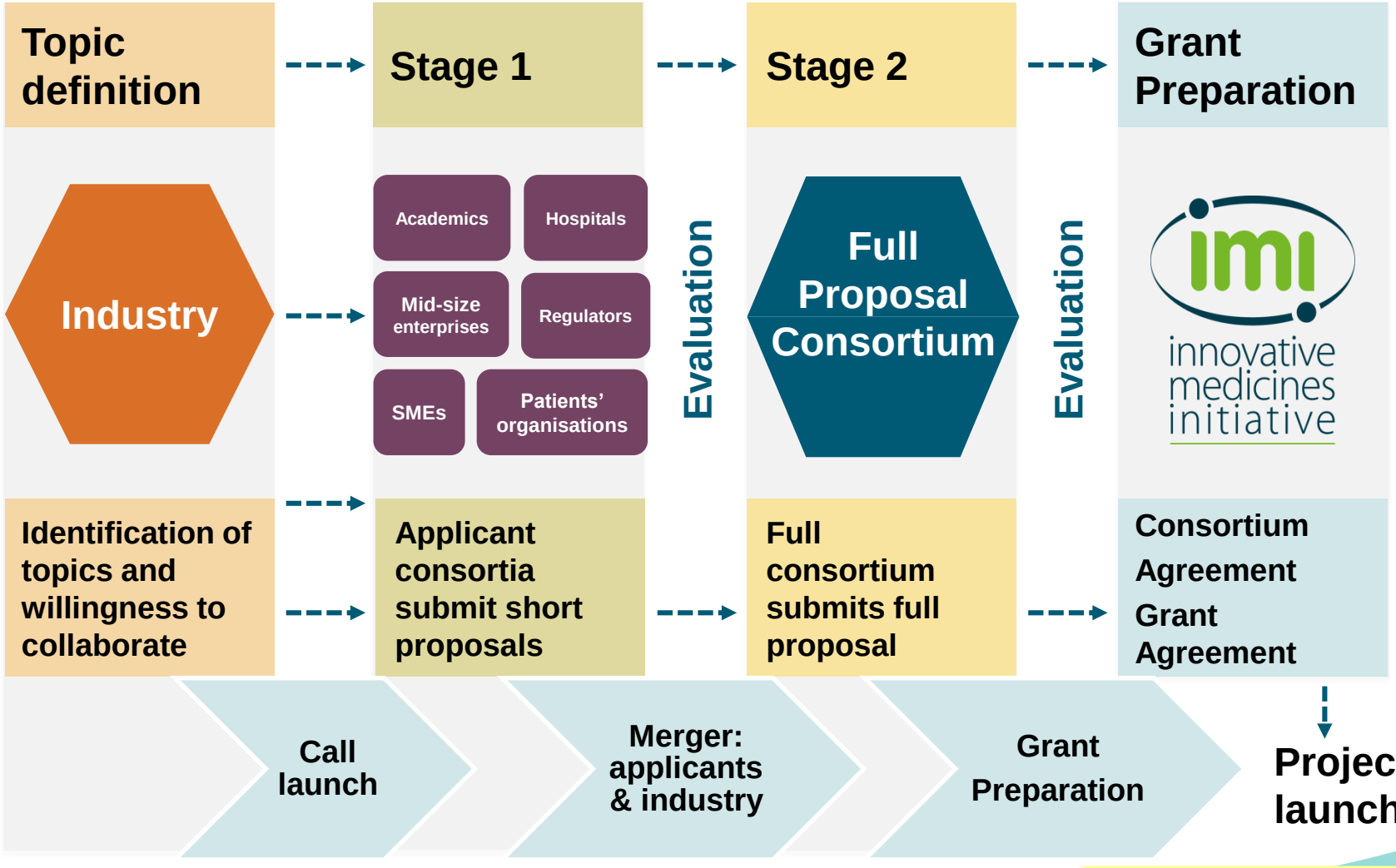
# Typical IMI project life cycle

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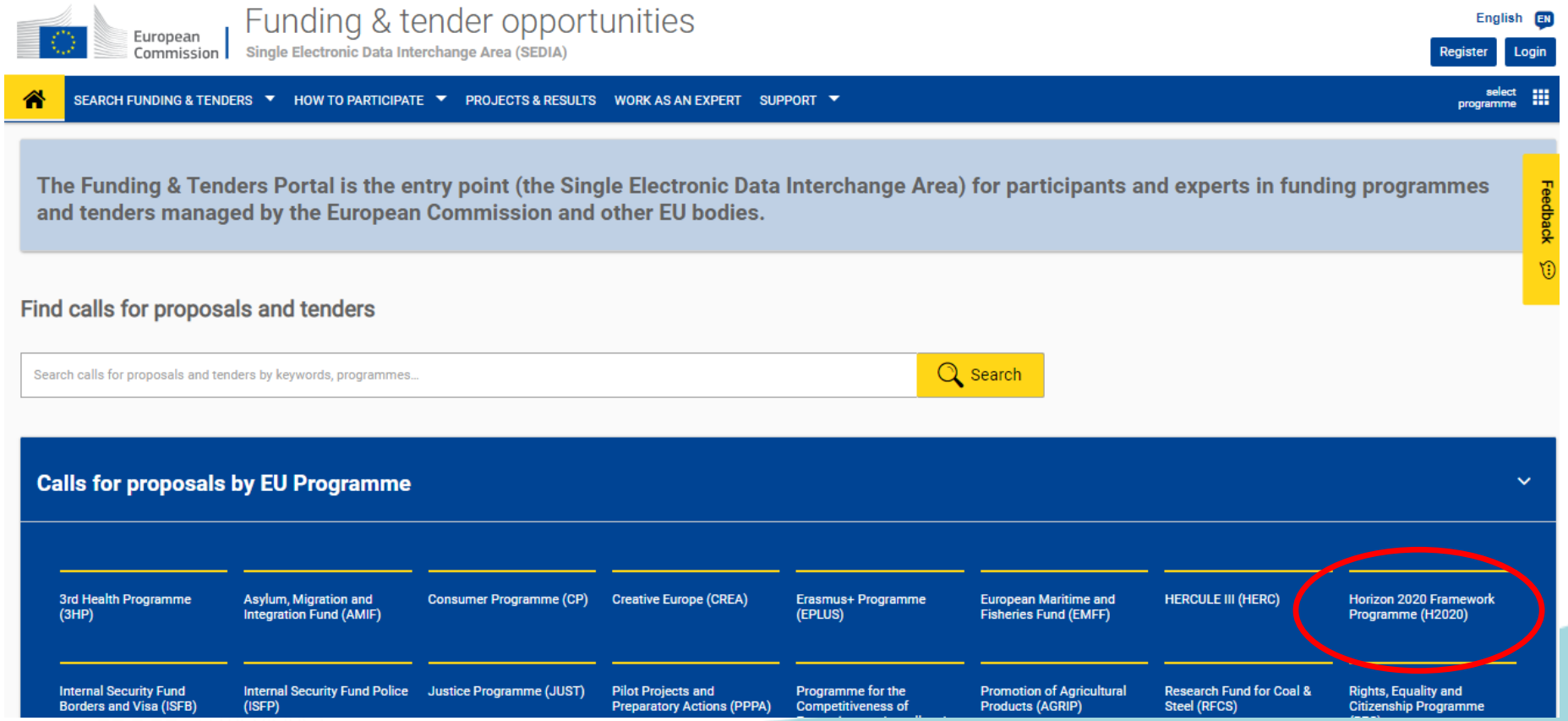
Expected GA signature – Summer 2021

# Submitting a proposal

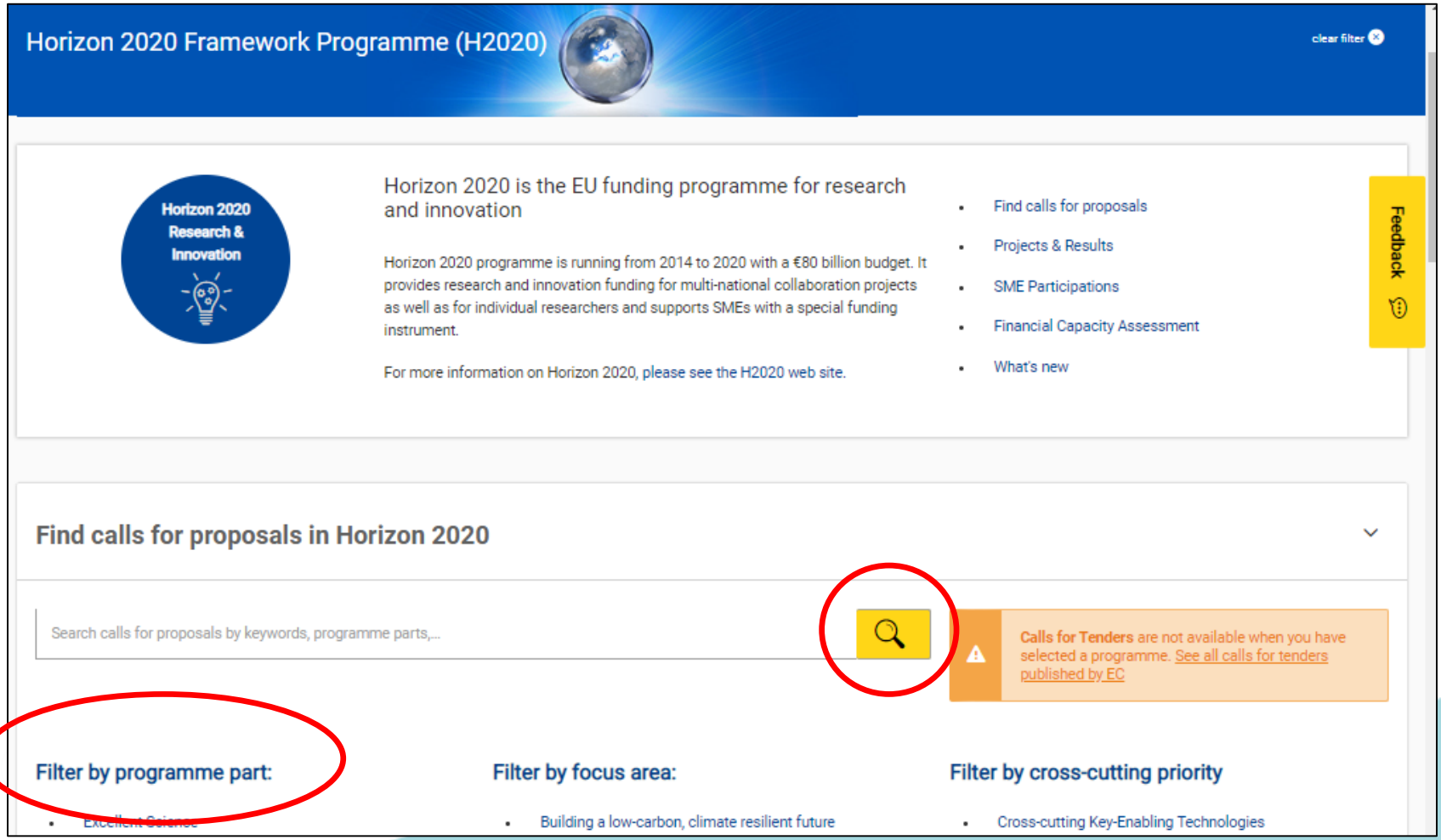
Via the **new** Funding and Tenders Portal

https://ec.europa.eu/info/funding-tenders/opportunities/portal/screen/home



# New Funding and Tenders Portal Horizon 2020 section

https://ec.europa.eu/info/funding-tenders/opportunities/portal/screen/programmes/h2020



# Proposal Template

- Available on IMI website & H2020 submission tool
- For first stage proposals, the page limit is **30 pages**.

**Title of Proposal**

**List of participants**

**Table of Contents**

**1. EXCELLENCE**

**1.1 Objectives**

**1.2 Concept and methodology**

**1.3 Ambition**

**2. IMPACT**

**2.1 Expected impacts**

**2.2 Outline Measures to maximise impact**

**3. IMPLEMENTATION**

**3.1 Outline of project work plan — Work packages, and major deliverables**

**3.2 Management structure and procedures**

**3.3 Consortium as a whole**

**3.4 List of work packages**

**4. PARTICIPANTS**

**4.1. Participants (applicants)**

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# Evaluation Criteria (1/2)

- **Excellence**
  - Level to which all the objectives of the Call topic text are addressed;
  - Soundness of the concept and credibility of the proposed methodology;
  - Extent that the proposed work is beyond the state of the art and demonstrates innovation potential;
  - Appropriate consideration of interdisciplinary approaches and use of stakeholder knowledge.
- **Impact**
  - Demonstration of how the outputs of the project will contribute to each of the expected impacts mentioned in the relevant Call topic text;
  - Outline of how the project plans to leverage the public-private partnership model to achieve greater impact on innovation within research and development, regulatory, clinical and healthcare practices, as relevant ;
  - Impacts on competitiveness and growth of companies including SMEs;
  - Quality of the proposed outline to:
    - Disseminate, exploit and sustain the project results;
    - Manage research data;
    - Communicate the project activities to relevant target audiences.

# Evaluation Criteria (2/2)

- **Quality and efficiency of the implementation**
  - Quality and effectiveness of the work plan outline, including extent to which the resources assigned to work packages are in line with their objectives and deliverables;
  - Appropriateness of the outline management structures and procedures;
  - Appropriateness of the allocation of tasks, ensuring that all participants have a valid role and adequate resources in the project to fulfil that role;
  - Complementarity of the participants and extent to which the consortium as whole brings together the necessary expertise;
  - Strategy to create a successful partnership with the industry consortium as mentioned in the Call topic text.

- 3 for each of the evaluation criteria ‘excellence’, ‘impact’ and ‘quality and efficiency of the implementation’
- the overall threshold is 10

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# Tips for writing a successful proposal

- Read **all the call-relevant material:**
  www.imi.europa.eu
- Begin forming your consortium **early**
  Partner search tools & networking events
- Provide **reviewers** with all the information requested to allow them to evaluate your proposal
- **Finalise and submit your proposal early**
- Contact the **IMI Office** (**NOT** industry topic writers):
  infodesk@imi.europa.eu

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# Common mistakes

- Admissibility/Eligibility criteria not met**:**
  - submission **deadline** missed
  - minimum of **3 legal entities** from **3 member states & H2020 associated countries** not met
- The proposal does **not address all the objectives** of the topic
- A proposal is **scientifically excellent** but will have **limited impact**
- **Complementarity** with Industry consortium not well described.

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# Find project partners

- Network with **your contacts**
- **Network** with fellow webinar participants
- Use **Partner Search Tools:**
  - EU Funding & Tenders portal: https://ec.europa.eu/info/funding-tenders/opportunities/portal/screen/how-to-participate/partner-search
  - German NCP partner search tool: www.imi-partnering.eu
- Get in touch with your **local IMI contact point**: www.imi.europa.eu/about-imi/governance/states-representatives-group
- Talk to your **Health National Contact Point** (NCP)
- Network on **social media** (e.g. IMI LinkedIn group)

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# Participation of SMEs, patient groups, regulators

We encourage the participation of a wide range of health research and drug development stakeholders in our projects.

- SMEs and mid-sized companies
- Patient organisations
- Regulatory bodies
- Companies / organisations from related fields (e.g. diagnostics, animal health, IT, imaging etc…)

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# StaRDust: Shortening the path to Rare Disease diagnosis by using newborn genetic screening and digital technologies

**Nicolas Garnier**

June 29th 2020 • IMI webinar

# Statement of the Issue

- ≈ 30 M Patients with a Rare Disease in EU. Less than 10% of Rare Disease Patients receive treatment and only 1% are managed using an approved treatment in EU

- Delivering effective treatments for Patients suffering from Rare Diseases has been described as one of the major global health challenges for the 21st century

- How might we design and execute a strategic program to address some of the major challenges faced by the Rare Disease Community, that can be endorsed and funded by IMI – EFPIA?

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# Rare Disease Patients face common problems:

- **Lack of access to correct diagnosis**
- **Delay in diagnosis**
- Lack of quality information on the disease
- Lack of scientific knowledge of the disease
- Inequities and difficulties in access to treatment and care

- Those can be summarized as follows:
  1. **Diagnosis**
  2. R&D New therapies
  3. Access

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# The Rare Disease conundrum

- Patients are not identified / diagnosed;
- Lack of epidemiological data;
- No natural history of disease data;
- No validated endpoint / patient-reported-outcomes (PROs);
- Patient are rare, experts are even rarer

- This has the pernicious additional effect of blunting interest in diagnosis / screening initiatives, as it would lead to patients being diagnosed with no concrete medical or clinical option
- This poses an ethical challenge, which unfortunately feeds the conundrum

# Overall objective of the full project

- ***Shorten the path to RD diagnosis by using newborn / paediatric (infants during their first weeks of life) genetic screening; and, via application of advanced digital technologies that enable rare disease diagnosis / identification. The latter might require consolidation of existing fragmented efforts.***

# Specific objectives

1. Assessment and **development of a comprehensive, strategic overview of existing converging RD resources** e.g. databases, registries, natural history projects, platforms, reference networks, rare disease academic centers of excellence, and initiatives for evaluation / identification of potential collaboration and synergies;
2. **Federation of available RD databases into a RD metadata repository amenable to machine learning** or other advanced digital tools;
3. **Co-creating a sustainable strategy for newborn genetic screening and pilot it**. This could start directly after achieving objective 1;

# Specific objectives (continued)

4. Based on the output of objectives 1 & 2:
   a. **Repurposing of pre-existing diagnosis AI algorithm to identify early onset RD patients in electronic health records**. This will include at least 3 pilots in better-known rare diseases where more robust data is available to train and test the AI algorithm(s), and / or;
   b. **Design and development of new AI algorithm(s)** to achieve the above goal.
5. Based on insights generated by objectives 1, 2 & 4, either repurposing or **development of a broad AI RD diagnosis "symptom checker"** to help undiagnosed RD patients going from one health care provider to another. In addition, exploration of further viable options to implement the symptom checker in actionable solutions for HCPs and patients.

# The Rare Disease Patient Diagnosis Odyssey

## Birth

- Sustainable strategy for newborn genetic screening

## Early Onset

- AI algorithm to "flag" Patients with better known Rare Disease in EHR

## HCP Cycling

- AI "digital clinical symptom checker"

**Strategic landscape analysis of converging initiatives / platforms & RD data sources**

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# Expected impact

- Early detection of rare genetic diseases would enable **early intervention** (when available), follow-up, and **genetic counselling** (such as family planning). This would result in **improved clinical and patient oriented outcomes**. Overall, this project will **increase public understanding around Rare Diseases**, and therefore **foster rare disease R&D**. A better understanding of rare diseases would also potentially lead to **better rare disease policies**, as well as **improved value-based healthcare**.

# Suggested architecture of the project

- It is suggested that each of the 5 objectives becomes a work package
- In addition, consideration should be given to a project management WP that would:
  - Ensure alignment between the participants as well as smooth internal and external communication;
  - Monitor compliance with the work plan;
  - Monitor planned resources and time schedule;
  - Coordinate fulfilment of all administrative milestones;
  - Ensure legal and data privacy requirements are met during the project lifetime

# Expected contributions of the applicants

- The consortium should include (but not limited to) the following key stakeholders:

- **Patient Organization**
- **Academia**
- **SMEs**
- **Public Health Decision Makers**
- **Regulators**

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# The consortium should mobilise the following expertise

- Objective 1:
  - Networking with EU, local Healthcare & Data Protection Regulators
  - Regulatory affairs, policy and politics, health economics, HTA / pharmaco-economy, regulatory sciences, legal / IP / licensing
  - Rare Disease expertise, international Dare Disease Patient Advocacy, Patient journey, public health,
  - Expertise in high & low-income EU health systems, public health systems Implementation.

# The consortium should mobilise the following expertise

- Objective 2, 4 & 5:
  - Data Exchange & Building Digital Infrastructure, User experience, Data security and Data Anonymisation, Methodology development, Data Management, Data Science, Data standards, Data translation
  - Pharmaco-epidemiology, Biostatistics, Bioinformatics, Software Engineering, Data stewardship, Business and governance model development (Including sustainability)
  - Medical, Legal General Data Protection Regulation (GDPR) Compliance, Data ethics, Privacy, Medical Insurance, Medical Training, Data Quality assurance, IT, Cyber security, Federated data

# The consortium should mobilise the following expertise

- Objective 3:

- Genetics, Genomics, Molecular Biology, Whole Exome and Whole genome Sequencing (WES / WGS)
- Gene panel, In silico panel
- Bioethics, Genetic Counseling

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# The consortium should mobilise the following expertise

- In general:

- Project Management, Study / Trial Operation Manager
- Medical / Scientific Writing
- Communications, Public Outreach

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# Expected contributions of public partners in the consortium

- Ideally, the consortium should welcome the participation of partners who could and would be willing to contribute:

- Rare Disease phenotypic data that could be integrated in the meta-data repository that would train the AI algorithm(s)
- Pre-developed rare disease recognition algorithm(s)

# Expected (in kind) contributions of industry consortium

- Project leadership and programme oversight, genetic research, medical affairs, data science / analytics & AI, epidemiology, regulatory, public relations / policy, commercial innovation;
- Scientific affairs, innovation, PPP management support, medical affairs, public affairs
- Genetic diseases / digital medical innovation, newborn screening, diagnostics, personalised medicine / healthcare, public policy, immunodeficiency

# Why join this endeavour ?

- Patients Advocacy Organizations
  - Paradigm change in rare disease diagnosis
  - Decreased time to the right diagnosis
  - Improved patient journey
  - Better healthcare

- Academia & SMEs
  - Advances in utilisation of digital technologies
  - Increased disease knowledge for future research
  - Improved data availability for future research

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# Why join this endeavour ?

- Public Health Decision Makers & Regulators

  - Implementation of digital transformation in healthcare
  - Improved diagnostic tools
  - Improved understanding of Rare Diseases
  - Higher accuracy in clinical decisions
  - Better care delivery
  - Increased trust in the healthcare system
  - Better use of data for public health
  - Improved value-based healthcare

# Key deliverables of the full project

1. Comprehensive landscape analysis of ongoing relevant initiatives and pre-existing resources with strategic recommendations
2. Federating of available RD databases into a RD metadata repository amenable to machine learning
3. RD gene panel for the purpose of NBS; list of criteria for inclusion / exclusion in the panel (scientific, technical, sustainable, and ethical) aligned with the overarching goals of action, with fully developed RD genetic New Born Screening protocol (and / or kit), tested and validated, with post-diagnosis planning recommendations, complemented by a proposed Whole Exome Sequencing / Whole Genome Sequencing approach
4. Digital diagnosis algorithms trained on the RD metadata repository to be used in electronic health records
5. Digital “clinical symptom checker” support tool trained on the federated RD database to help RD patients cycling through HCPs

# Thank you

**All questions** should go through the **IMI Executive Office**

www.imi.europa.eu

@IMI_JU

# Involvement of SMEs, patient groups, regulators

# SME participation

IMI encourages the participation of SMEs in applicant consortia as they can offer a complementary perspective to other organisations.

Contribution of SMEs would be considered especially beneficial in providing the following expertise and activities :

- data curation, harmonisation, interoperability
- bioinformatics, diagnostic algorithm development and AI applications
- health economics
- public-health, public relations and communication
- project management

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# Patient participation

Involvement of patient organisations is imperative for this topic to ensure patient centricity and patient oriented outcomes

In particular your patient partners input would be of importance on:

- discussions on feasibility and acceptability of proposed new born or infant screening
- definition of the RD gene panel for newborn screening
- ethics, data privacy and health policy
- aspects of post-diagnosis planning recommendations (genetic counselling, referral, etc.)
- health economics, outcomes and benefits for patients and families.
- community outreach and dissemination

***"The patient, doctor and researcher – each is a different kind of expert."***

# Interactions with regulators

- **Have a plan for interaction** with relevant **milestones** and **resources** allocated, as needed
- Consider the **formal regulatory process** to ensure **regulatory acceptance of project results** (e.g. qualification procedure for biomarkers)
- Get familiar with **services offered for dialogue** (e.g. at EMA through qualification advice, Innovation Task Force, briefing meetings)
- Consider involving regulators as project participants or in the **advisory board**
- Have a plan for dialogue with **HTA bodies / payers**, if relevant

**To maximise impact of science generated by projects**

**Engage in dialogue with regulatory authorities**

More info:

- Webinar & presentations 'How to engage with regulators EMA / FDA'
- 'Raising awareness of regulatory requirements: A guidance tool for researchers'

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# Thank you

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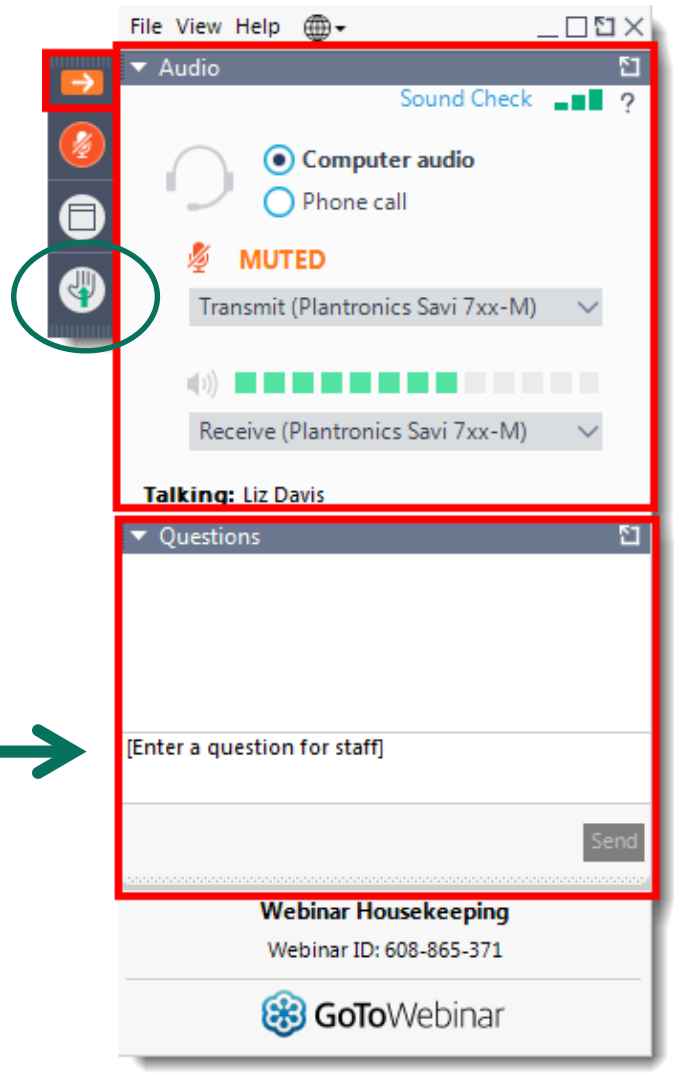
@IMI_JU

# Questions & answers

# Questions?

Raise your hand if you want to ask a question orally




Send a question in writing

After the webinar, send any questions to the **IMI Programme Office**

applicants@imi.europa.eu

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# Thank you!