IMI2 – Call 23: Shortening the path to **rare disease diagnosis** by using new born genetic screening and digital technologies
Potential contribution of the Izmir Biomedicine and Genome Center (IBG, Turkey) to rare disease diagnosis
Keywords: rare diseases, genetic screening, curated patient data, patient samples, biobanking

1. About IBG: The Izmir Biomedicine and Genome Center (IBG, www.ibg.edu.tr) is a recently established (2014) advanced biomedical research and innovation centre with modern, state-of-the-art infrastructure and facilities, which are exceptional in Turkey in regard to quality, size, and their unique composition. Furthermore, IBG is one of four national Centres of Excellence and the only one in the biomedical field, making IBG the prime address for biomedical research in Turkey. It currently hosts 27 basic, translational, and technological research groups that work on multiple aspects of biomedicine and genome sciences. This multidisciplinary, cooperative nature is seen as a strong asset to address complex scientific questions.

2. IBG & rare diseases: A major research focus of IBG are rare diseases, which are approx. twice as prevalent in Turkey than in other ERA countries. IBG established a wide network of relevant stakeholders ('Quadruple Helix' approach) and is the national node for several international rare disease research consortia (e.g. CONSEQUITUR, ICGNMD) with partners from over a dozen countries (e.g. Broad Inst., Cambridge U., CNAG-CRG, MIT, Newcastle U., Ottawa U.). Our efforts in this field were recently recognized by the EC by awarding IBG the prestigious ERA Chairs grant RareBoost (scheduled to start autumn 2020). This ERA Chairs project will greatly facilitate the development of IBG's rare diseases focus towards an international level of excellence in research and innovation.

3. What we can offer: IBG would like to contribute its expertise, infrastructure, and national network to rare disease research, including the IMI2 call 23 on rare disease diagnosis. We are looking to join a consortium as WP leader or contributor, depending on its needs. In the context of this call, IBG's following assets might be of interest:

- Expertise: IBG has several ongoing multidisciplinary research programs on rare diseases, with a current focus on multi-tiered genetic approaches, including comprehensive genetic analysis of patients with rare diseases. With their strong genetic/epigenetic, molecular, in silico, bioinformatic, and computational expertise, IBG's researchers are able to generate, analyse, and model 'omics-data' and interconnect them with other databanks. The complete methodological portfolio for a mechanistic investigation is established, including cell biological (e.g. lab-/organ-on-a-chip culture techniques), multi-omic, and *in vivo* (transgenic mouse models) approaches. Furthermore, we demonstrated our ability to translate research findings to innovation. For example, diagnostic kits developed by IBG researchers for the screening of three rare metabolic diseases in neonates were already adopted into the national standard of care (approx. 1.5 million tests/year). Moreover, IBG members repeatedly advice national policy-makers on issues related to health, rare diseases, genome science, and biotechnology.
- Curated patient samples and data: IBG offers an established pipeline for collecting phenotypic data in a standardized way (AI applicable), deep phenotyping, as well as the mechanistic analysis and the effective integration with international collaborators. Within our ERA Chairs project, IBG plans to spearhead and coordinate a national rare disease registry and repository including curated data (detailed and standardized) and patient material (biobank). By establishing itself as the national hub for rare disease research and innovation, IBG will be able to coordinate and moderate interactions with clinicians all over Turkey and MENA countries.
- Infrastructure: IBG is a national rare disease research centre of excellence whose state-of-the-art research and translational facilities cover the TRLs 1-7. Its biobank is the national node for BBMRI-ERIC. IBG is located on the Health Campus of Dokuz Eylul University and several of the university's clinicians, attending rare disease and paediatric patients, are affiliated to IBG.

4. Contact: For more details and for brainstorming on potential contributions please contact Yavuz Oktay (yavuz.oktay@ibg.edu.tr) or Gerhard Wingender (gerhard.wingender@ibg.edu.tr).