GEMS-APP

A WEB-BASED DYNAMIC CONSENT APPLICATION FOR STUDYING GENOME-WIDE EPISTASIS IN MARFAN SYNDROME

Eneko Urizar PhD, Thibaut You-Hout, Harold Smits, Nicolas Callebaut, Florent Dubois, Bruno Fonteyn, Thomas Dubuisson LLM, Nassim Versbraegen PhD, Tom Lenaerts PhD, Guillaume Smits MD PhD, Angelika Jürgens, Paul J. Coucke PhD, Lotte J. F. Van Den Heuvel, Guillaume Jondeau MD PhD, Bart L. Loeys MD PhD, Ludivine Verboogen LLM and Romain Alderweireldt LLM,

for the Marfan Project of the 101 Genomes Foundation www.f101g.org

182 Consents | **93** Days | **20** Countries | **147** *FBN1* | **29** Ile2585Thr | **1/3** Pheno

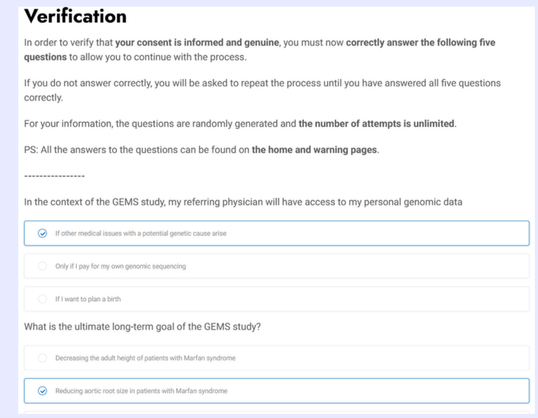
BACKGROUND

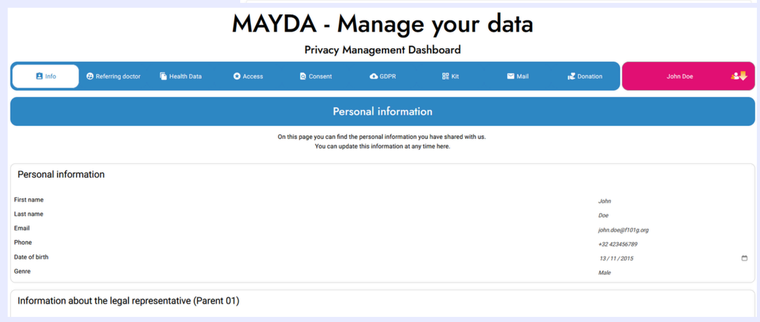
The challenge. Rare disease genomics requires large cohorts and faces barriers in informed consent, participant engagement, and regulatory compliance.

The opportunity. Electronic dynamic consent allows patients to manage their participation over time while supporting long-term genomic research.

Our approach. In Marfan syndrome, where identical *FBN1* variants can produce highly variable cardiovascular outcomes, we have developed GEMS-App, a secure compliant web platform enabling dynamic consent and remote participation. Join GEMS at **www.101gems.be**

GEMS & MAYDA





MAIN FINDINGS

Figure 1. GEMS-App accommodates all time frames

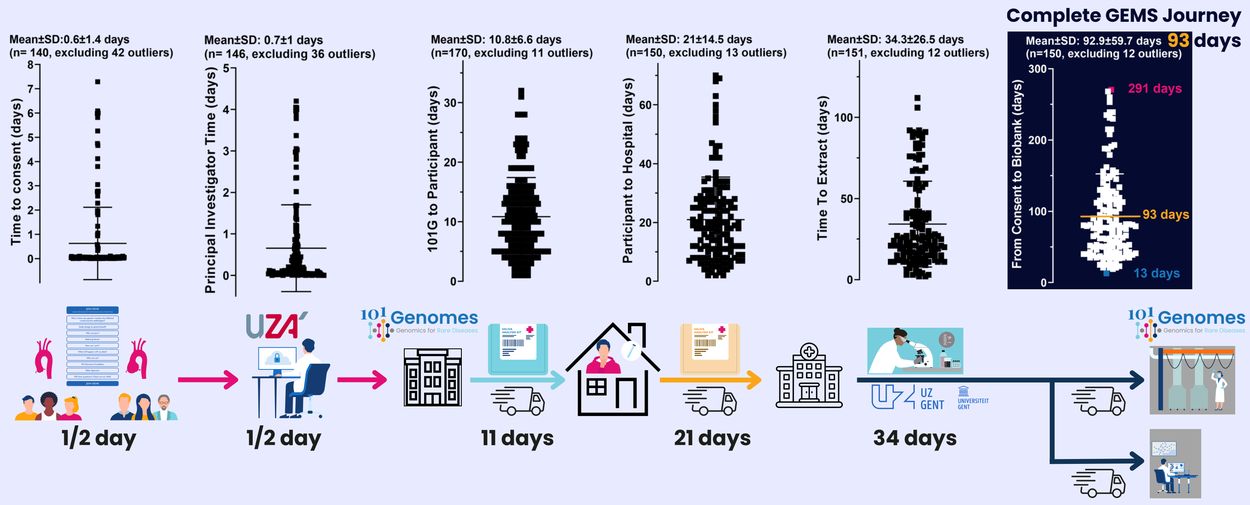
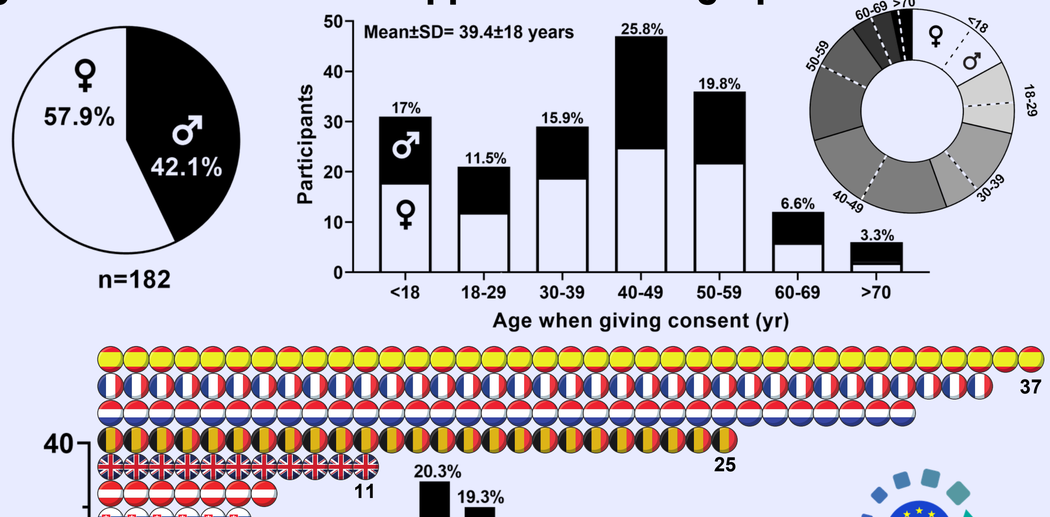


Figure 2. First F101G GEMS-App cohort demographics



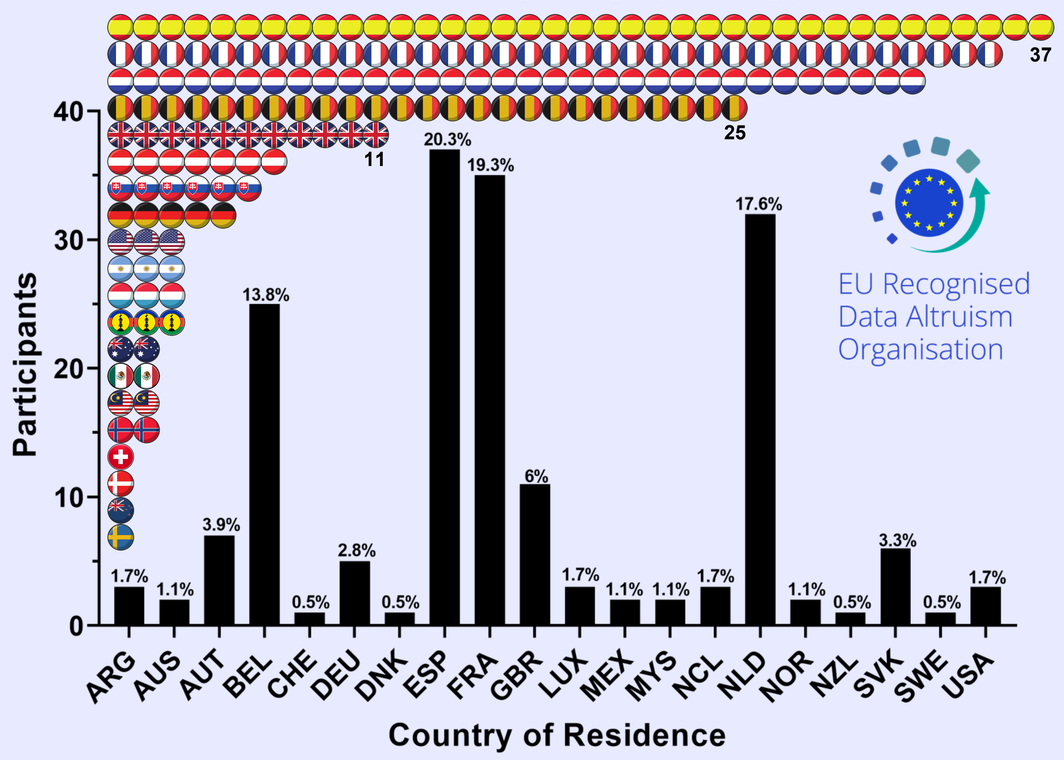
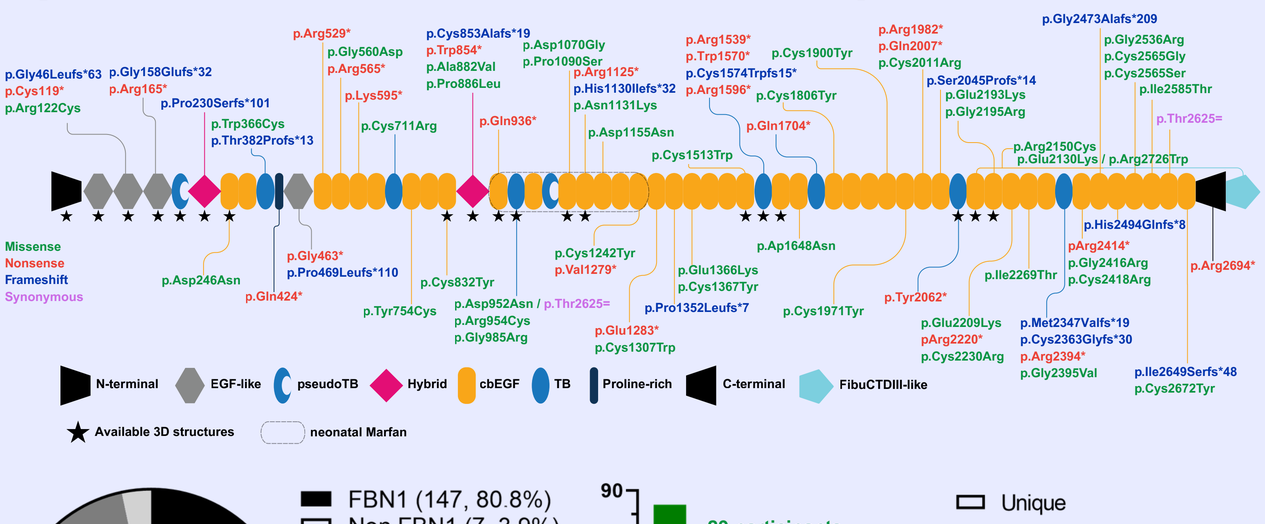
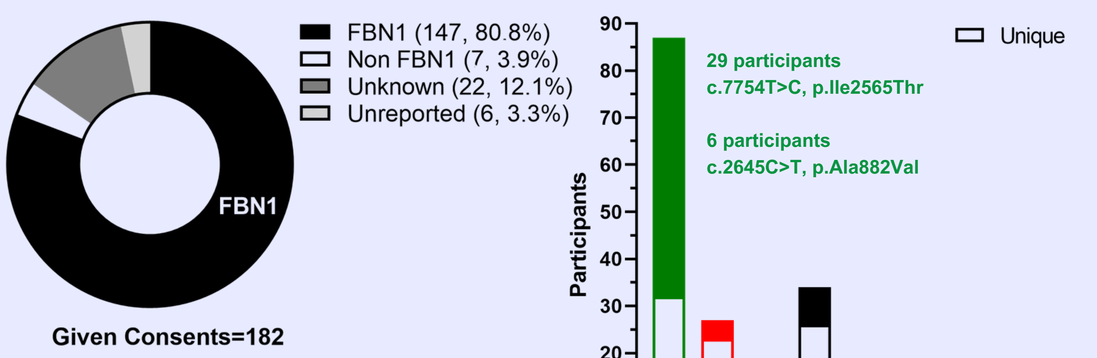


Figure 3. Variants across the entire Marfan causing FBN1





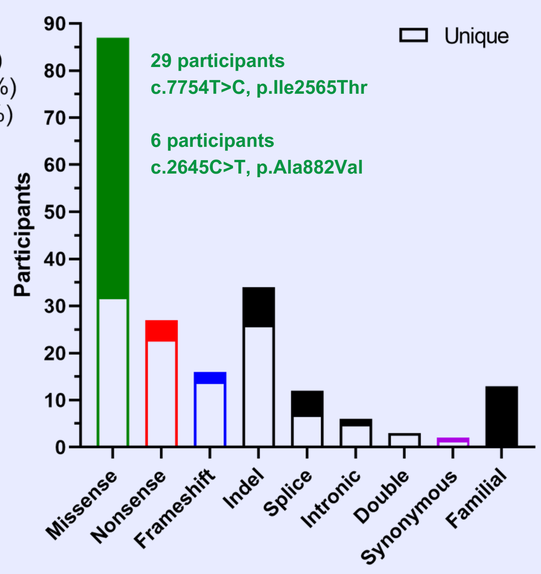
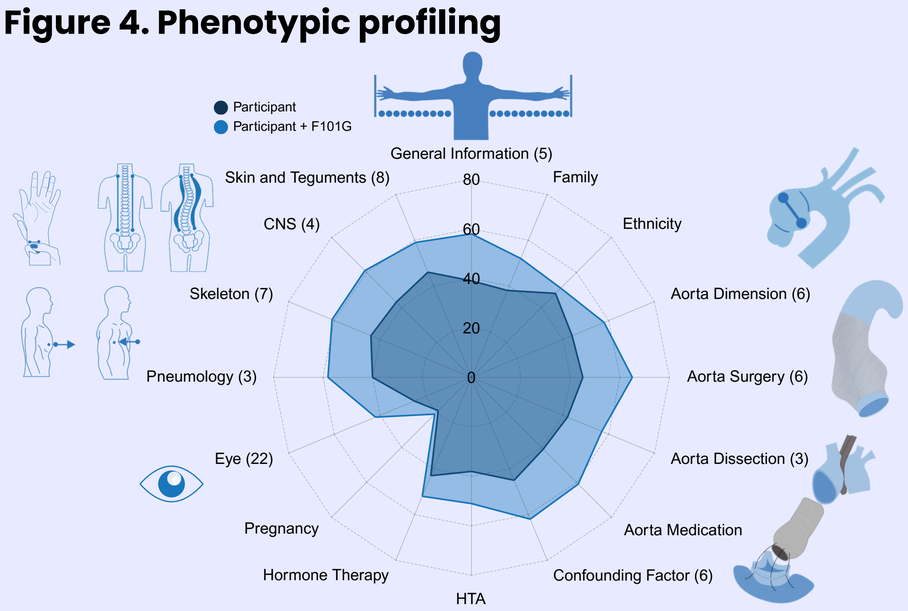


Figure 4. Phenotypic profiling



CONCLUSIONS

Dynamic consent can be implemented securely, legally, and at scale in rare disease genomics. GEMS-App enables remote flexible and empowered recruitment, participant-controlled data sharing, and global cohort building for Marfan syndrome, providing a scalable framework for future rare disease research.