

101 Genomes

Genomics for Rare Diseases



GEMS-App: A Web-Based Dynamic Consent Platform Enabling Genome-Wide Epistasis Research in Marfan Syndrome

ECRD 2026

3rd June 2026 – Prague

Romain Alderweireldt

- **CMS | LLM - Competition law** Telecommunication & Life sciences
- **Belgian Marfan Syndrome Association (ABSM)** | Board member
- **Genome4Brussels | Consortium** ULB Machine Learning Group, IB2/HUB Center of Human Genetics and 'fair genomics': Development of monogenic, polygenic and oligogenic genome analysis tools based on AI
- **Marfan Europe | Scientific WG** - Head
- **ERN | VASCERN | HTAD** group
- **ERN | ELSI** Legal & Ethical Issues and relations with Stakeholders inter-ERN transversal Working Group (composed of representatives of the Member State and the EU Commission) in charge of legal and ethical issues faced by European Reference Network on Rare Multisystemic Vascular Diseases.
- **1+MG & B1MG | 1+ Million Genomes & Beyond 1 Million Genomes**
<https://b1mg-project.eu/> Member of the Belgian Mirror Group for WP02/WG02 ELSI and WP07/WG08 Rare diseases
- **101 Genomes Foundation | Co-Founder** Genomics for Rare Diseases



Ludivine & Romain ALDERWEIRELDT-VERBOOGEN

BACKGROUND

Marfan syndrome



Marfan syndrome

- Eye problems
- Abnormal chest, heart and lung problems
- Short torso
- Tall, thin body frame
- Long arms, legs and fingers

A

Chromosome 15

B

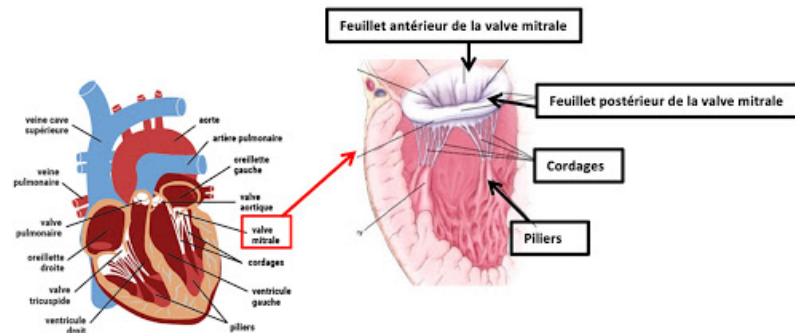
FBN1 Gene, 237.5 Kb, 65 Exons

Neonatal Marfan



A very different story...

- These are almost always spontaneous cases: **de novo**
- Genetic analysis reports that these cases are usually (but not always) found when a pathogenic mutation occurs in the core of the FBN1 gene on **the interval of exons 24 to 32**
- A signature of this form is the rapid affection of the **mitral valve**



Exons 24 - 32 on FBN1

« Marfan syndrome (MFS) (OMIM 154700) is an autosomal dominant disorder of fibrous connective tissue involving the ocular, skeletal, and cardiovascular systems. MFS patients present with clinical variability, in which the rare neonatal Marfan syndrome (nMFS) has the most severe presentation in early childhood. The prognosis of nMFS is very poor, **with a mean survival age of only 16.3 months**. Valvular insufficiencies and diaphragmatic hernias have been associated with shorter survival in patients diagnosed before the age of 1 year. [...] The term neonatal Marfan syndrome was first used in 1991 to describe the most severe phenotype of MFS similar to cases previously known as infantile Marfan syndrome, congenital Marfan syndrome, and severe perinatal Marfan syndrome. Recently, it has been suggested that the term neonatal MFS should be replaced by **early onset and rapidly progressive MFS** to represent the **most severe features of MFS in early childhood** ».

PENG Q. et al., « A novel fibrillin-1 gene missense mutation associated with neonatal Marfan syndrome : a case report and review of the mutation spectrum », BMC Pediatrics, 30 avril 2016, 16:60, DOI 10.1186/s12887-016-0598-6

PARTICIPANTS NEEDED FOR NEONATAL MARFAN SYNDROME STUDY



Shaine Morris, MD
 Associate Professor of Pediatrics -
 Cardiology
 Texas Children's Hospital

Send message

View bio

For the Love of Neonatal Marfans

Groupe Privé · 72 membres



+ Inviter

Quid for FBN1?



MARFAN

The UMD-FBN1 mutations database

www.umd.be/FBN1/
3000 variants

CONTROLES
(Not MARFAN)

gnomAD

genome aggregation database

gnomAD v2.1.1 Search by gene, region, or variant

<https://gnomad.broadinstitute.org/>
180.000 variants

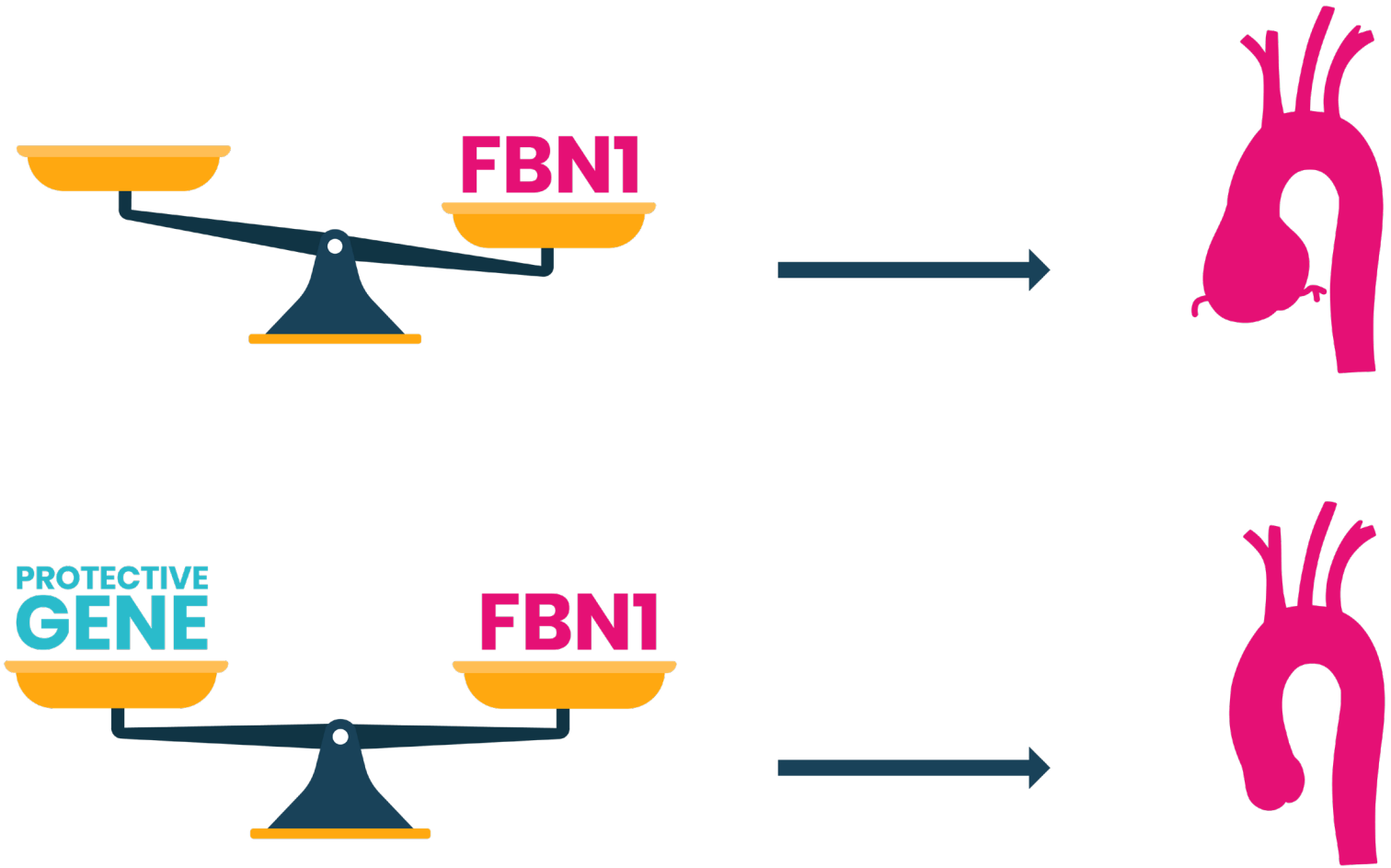


Discovery **at the intersection of 122 individuals in gnomAD** carrying a mutation on the FBN1 gene identified in the literature as **pathogenic** and causing Marfan syndrome.

Among these 122 individuals, **24 individuals have mutations located in the area of exons 24 to 32 where neonatal forms of Marfan syndrome are found.**

This observation was subsequently confirmed in the scientific literature in 2019. See BAUDHUIN L. ET AL., « Variability in gene-based knowledge impacts variant classification: an analysis of FBN1 missense variants in ClinVar », *EJHG*, 21 May 2019, <https://doi.org/10.1038/s41431-019-0440-3>.

Modifier genes



The quest for protective genes: GEMS design



Most common missense variant: *FBN1*
p.Ile2585Thr
c.7754T>C

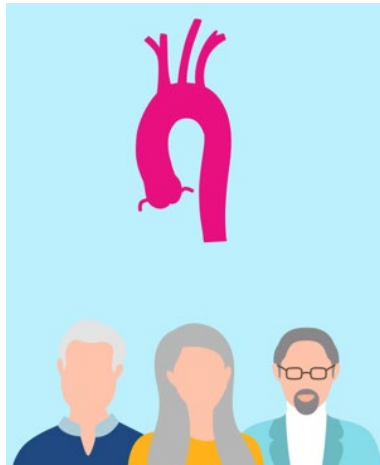
Cohort: > 200 mutation carriers assembled

GnomAD: asymptomatic cases

Clinical phenotyping: extreme variability demonstrated

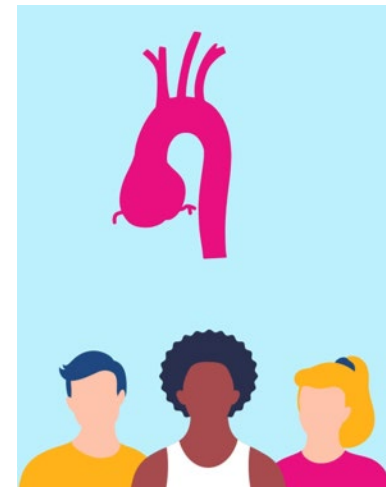


Normal



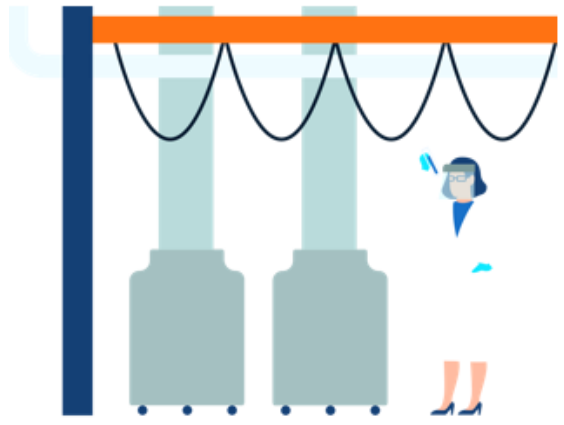
UMC
Unaffected Mutation Carrier

Severe



AMC
Affected Mutation Carrier

Biological data → 101 Biobank



Cliniques universitaires de Bruxelles
Route de Lenak, 80B
B-1070 Bruxelles
T: +32 (0)2 555 31 11
M: contact@erasme.ulb.ac.be
S: www.erasme.ulb.ac.be



Dr VERBOOGEN Michel
Médecin, Neuropsychiatre,
Président du Conseil d'Administration
de la Fondation 101 Génomes
Rue Cambion, 31
7500 Tournai

Bruxelles, le 05/04/2022

Comité d'Ethique
hospitalo-facultaire
Erasme - ULB
(021 / 406)

Cher VERBOOGEN Michel,

Dans le cadre de sa mission légale définie à l'article 7, §1^{er}, alinéa 3, de la loi du 19 décembre 2008 relative à l'obtention et à l'utilisation de matériel corporel humain destiné à des applications médicales humaines ou à des fins de recherche scientifique, le Comité d'Ethique hospitalo-facultaire Erasme-ULB a pris connaissance du dossier de soumission de la biobanque de MCH « Biobanque de matériel corporel humain de la Fondation 101 Génomes » (Ref. B2022/001).

Le dossier évalué comprend les pièces suivantes :

- > Cover Letter, dated March 13, 2022
- > Demande d'avis du Comité d'Ethique, dated March 13, 2022
- > Annexe - lien : 20220313 F101G Biobanque

Le Comité octroie, en date du 05/04/2022, un avis favorable par rapport aux activités et objectifs, tels que décrits dans le dossier de soumission

Nous vous prions d'agréer, Cher



Agence fédérale des médicaments et des produits de santé
Avenue Gallée 3/3
1210 Bruxelles
www.afmps.be

Cellule Matériel Corporel Humain
Philippe De Buck
Tél. : +32 2 528 40 00
e-mail : biobanks@fagg-afmps.be

FIOIG Biobanque
Avenue de Sumatra 6
1150 Ixelles
Belgique

Votre lettre du	Vos références	Nos références	Annexe(s)	Date
		0000	0	

Objet: Notification établissement biobanque

Madame, monsieur,

J'accuse bonne réception de votre notification concernant votre biobanque conformément à l'art. 3 de l'arrêté royal du 9 janvier 2018 relatif aux biobanques. Nous avons reçu la notification en date du 31-05-22 et nous vous confirmons que le dossier est complet et recevable.

En date du 09-06-2022 le numéro de notification **BB220008** est accordé à la biobanque sise à FIOIG Biobanque Avenue de Sumatra 6 1150 Belgique

L'exploitant de la biobanque est : **Fondation 101 Génomes**
Le gestionnaire du matériel corporel humain au sein de la biobanque est : **Verboogen, Michel**

Ce numéro de notification vous est accordé sans préjudice de toutes consultations ou vérifications ultérieures relatives à la conformité aux dispositions de l'arrêté Royal 9 janvier 2018 relatif aux biobanques.

Toute modification aux renseignements fournis la pour la présente notification ou toute cessation temporaire ou définitive envisagée des activités de la biobanque doit, conformément à l'art. 4 l'arrêté Royal du 9 janvier 2018, immédiatement être signalée à l'Agence Fédérale des Médicaments et des Produits de Santé. Ces informations peuvent être communiquées soit par lettre soit par courriel adressée à l'adresse biobanks@fagg-afmps.be, toujours en mentionnant le numéro de notification.

Veillez agréer, Madame, Monsieur, l'expression de nos salutations distinguées,

afmps fagg
Digitally signed by
Philippe De Buck
(Signature)
Date: 2022.06.10
17:41:24 +02'00'

Philippe De Buck,
Chef de division autorisation.

Digital data → 101 Genomic Cloud



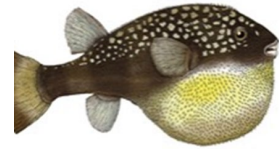
Humanizing Genomics
macrogen
EUROPE

First ever interconnection between macrogen and azure

101
Genomic Cloud

Azure

Genomics in the Cloud
Using Docker, GATK, and WDL in Terra



Geraldine A. Van der Auwera & Brian D. O'Connor

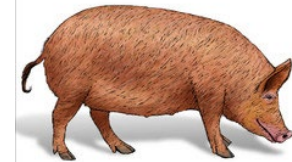


Dr Geraldine Van der Auwera

Global Alliance for Genomics & Health (GA4GH)

Broad Institute of MIT and Harvard

O'REILLY
Genomics in the Azure Cloud
Scaling Your Bioinformatics Workloads Using Enterprise-Grade Solutions



Colby T. Ford



Dr Colby T Ford

Cloud AI and Genomics architect

Classic process



Suboptimal process

Principal investigator



1X
Ethics Committee

2X
Ethics Committee



Doctor



Potential participant

?



?

Informed Consent
(Indirect & static)

GEMS App process

Direct interaction,
Objectivation of consent,
Effective exercise of the rights

Principal investigator



1X
Ethics Committee

MAYDA
Manage your data

GEMSAPP

Informed Consent
(Direct & dynamic)



Ethics ~~Committee~~



Doctor



Potential participant

?



?

Informed Consent
(Indirect & static)

GEMS & MAYDA

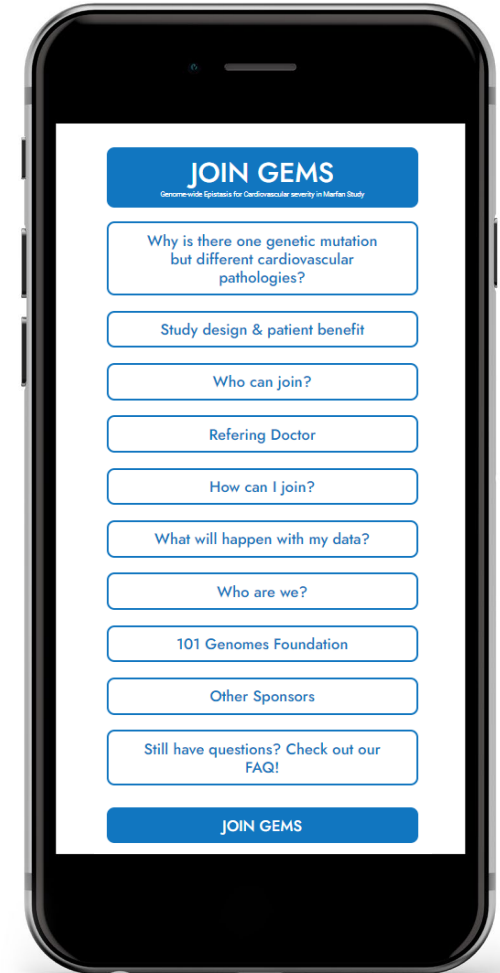
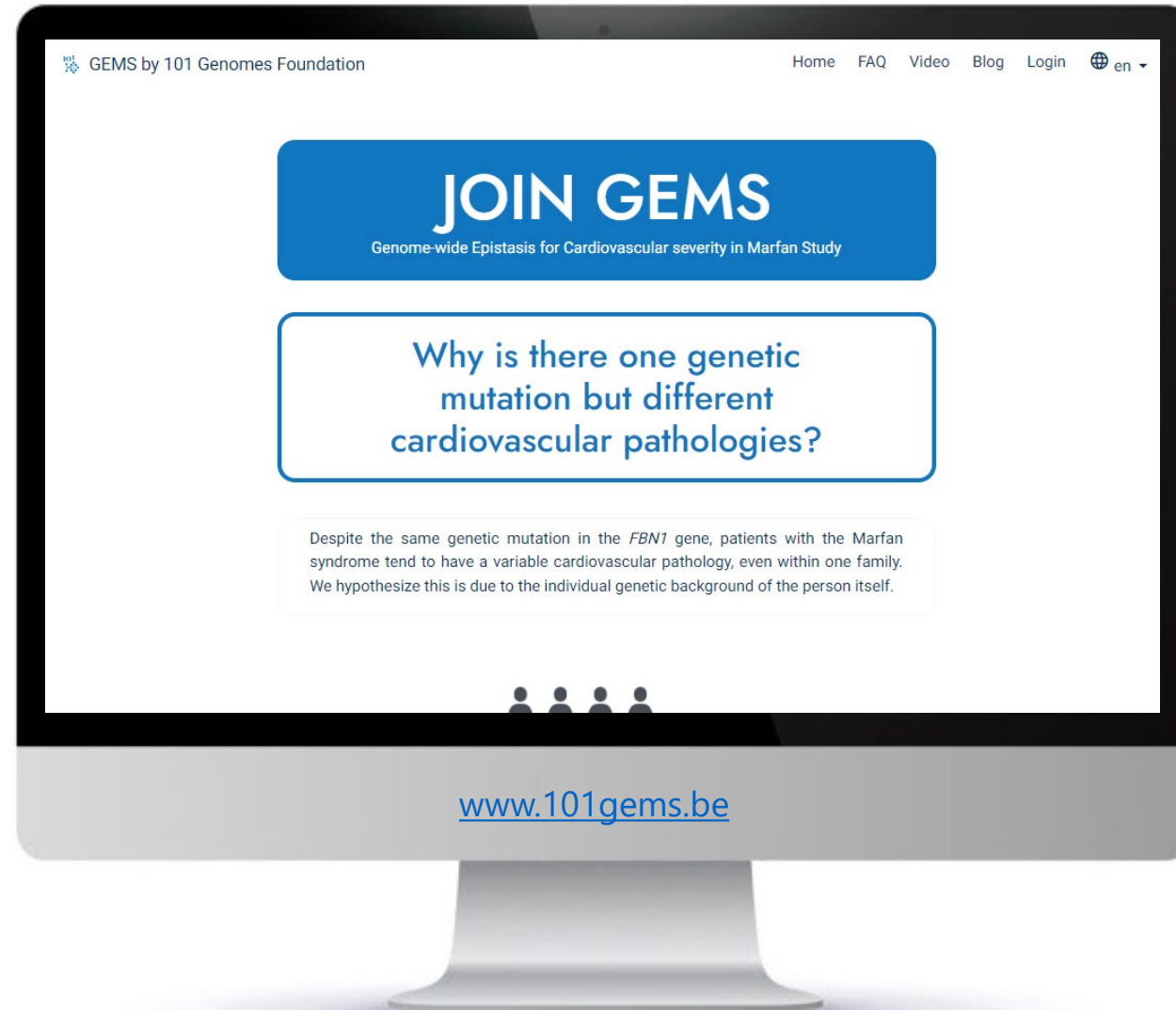
GEMS App



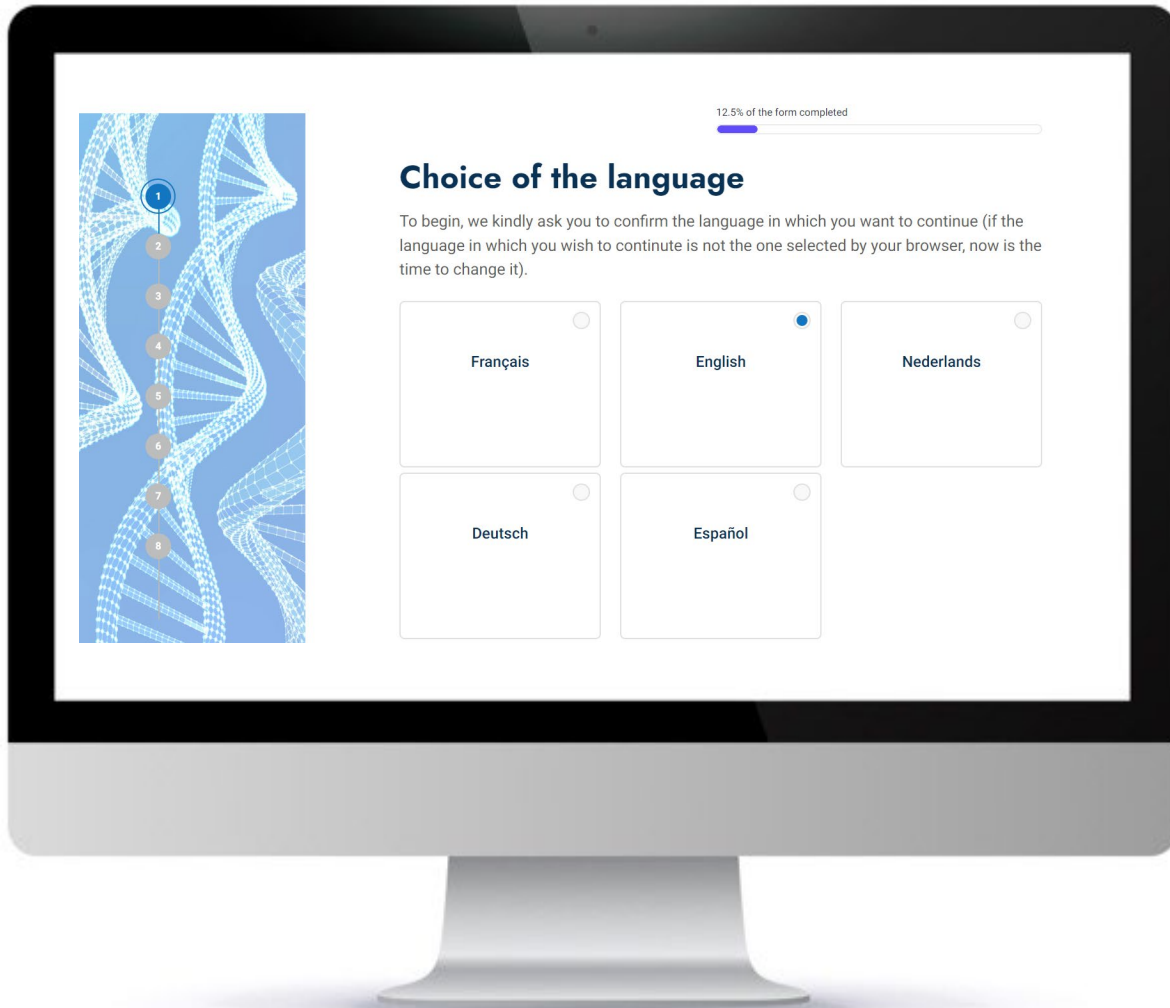
Web application for

- consent collection,
- consent management and
- phenotypic data collection

<https://www.101gems.be>



Multilingual and operational



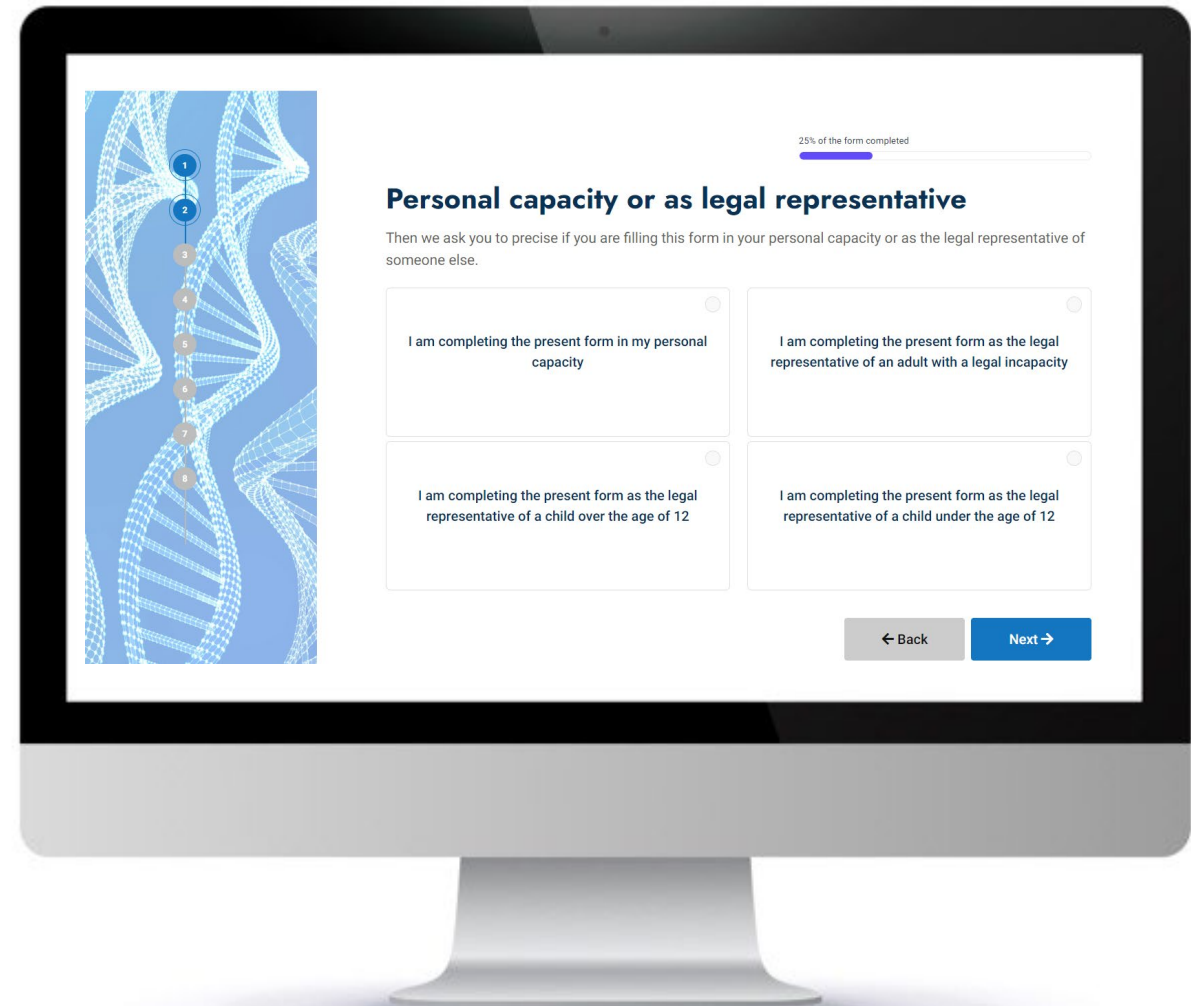
12.5% of the form completed

Choice of the language

To begin, we kindly ask you to confirm the language in which you want to continue (if the language in which you wish to continue is not the one selected by your browser, now is the time to change it).

Français
 English
 Nederlands

Deutsch
 Español



25% of the form completed

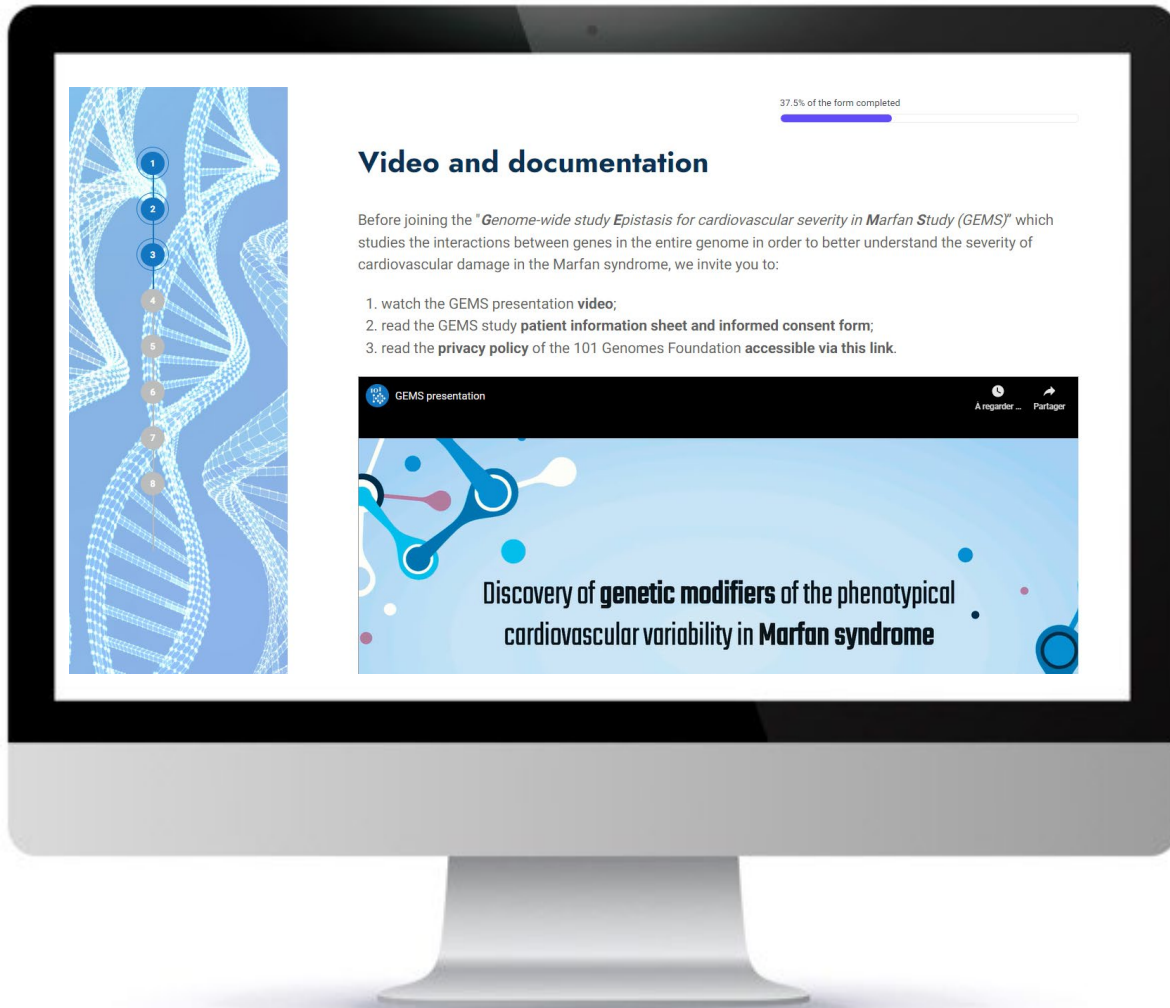
Personal capacity or as legal representative

Then we ask you to precise if you are filling this form in your personal capacity or as the legal representative of someone else.

I am completing the present form in my personal capacity
 I am completing the present form as the legal representative of an adult with a legal incapacity

I am completing the present form as the legal representative of a child over the age of 12
 I am completing the present form as the legal representative of a child under the age of 12

Full process and verification

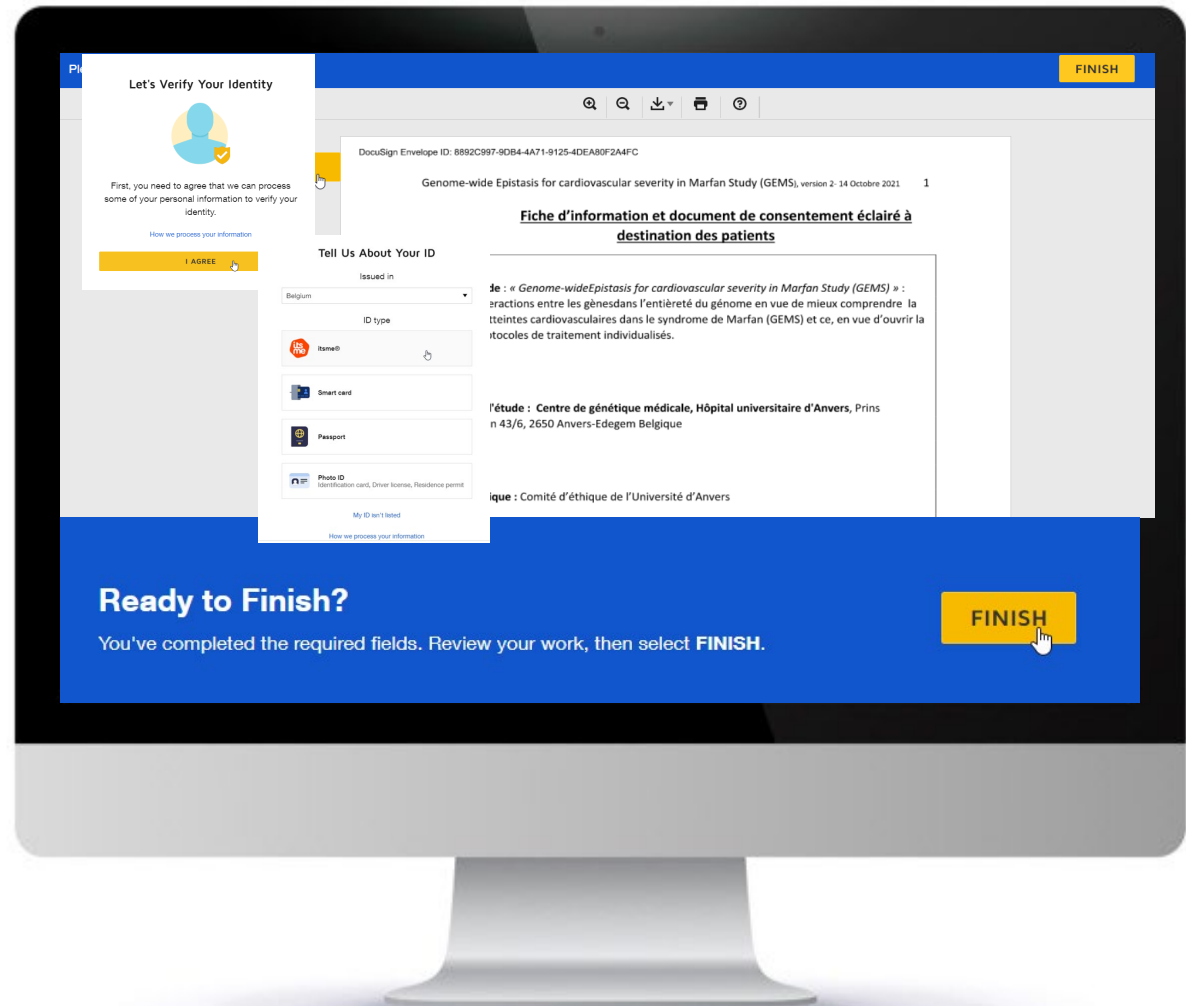


Consent directly with the principal investigator

GEMS consent

INFORMED CONSENT: Investigation of genome-wide gene interactions to better understand cardiovascular severity in Marfan syndrome to pave the way for individualized treatment protocols.

- I confirm that I am thoroughly informed about this study and received a copy of the "patient information sheet and informed consent".
- I read these documents, understood this information and had sufficient time to ask questions. My research physician has explained the study with regards to the conditions, duration, the effect and risks.
- I understood that my participation in this study can be stopped any time after informing my research physician, without any influence for further medical care.
- I give permission to the initiator of this study, Prof. Dr. Bart Loeys and associated organisations (after pseudonymisation) to have access to my medical records. My medical records will be handled strictly confidential and used in the framework of this study.
- I give permission to assembly, process and use my medical records as described by the information sheet for the patient. I also give permission to use and process this data in other European countries within the international context of this study.
- I voluntarily give my consent to participate in this research and all associated examinations. I am willing to give further information about my medical history, use of medications, and cooperation in other studies.



The screenshot shows a web interface for completing the consent form. On the left, a 'Let's Verify Your Identity' panel includes a 'I AGREE' button. The main area displays a DocuSign envelope for 'Genome-wide Epistasis for cardiovascular severity in Marfan Study (GEMS), version 2 - 14 Octobre 2021'. Below this, there is a section titled 'Fiche d'information et document de consentement éclairé à destination des patients' containing study details in French. At the bottom, a blue banner asks 'Ready to Finish?' and provides instructions to review work and click the 'FINISH' button.

Let's Verify Your Identity

First, you need to agree that we can process some of your personal information to verify your identity.

How we process your information

I AGREE

Tell Us About Your ID

Issued in: Belgium

ID type:

- NamID
- Smart card
- Passport
- Photo ID (Identification card, Driver license, Residence permit)

My ID isn't listed

How we process your information

DocuSign Envelope ID: 8892C997-9DB4-4A71-9125-4DEA80F2A4FC

Genome-wide Epistasis for cardiovascular severity in Marfan Study (GEMS), version 2 - 14 Octobre 2021 1

Fiche d'information et document de consentement éclairé à destination des patients

Je : « Genome-wide Epistasis for cardiovascular severity in Marfan Study (GEMS) » : interactions entre les gènes dans l'entière du génome en vue de mieux comprendre les atteintes cardiovasculaires dans le syndrome de Marfan (GEMS) et ce, en vue d'ouvrir les protocoles de traitement individualisés.

l'étude : Centre de génétique médicale, Hôpital universitaire d'Anvers, Prins n 43/6, 2650 Anvers-Edegem Belgique

lique : Comité d'éthique de l'Université d'Anvers

Ready to Finish?

You've completed the required fields. Review your work, then select **FINISH**.

FINISH



DNAgenotek Oragene® 600 series product weight and dimensions

Catalogue numbers	Saliva volume	Pre-collection			Post-collection	
		Package dimensions and contents — WEIGHT 34-38g†			Total sample volume††	Tube dimensions (mm)
		Clamshell (mm)	Components	Collector		
Without 2D bottom barcode	OG00-600	2 mL	 Instructions for use Cap	 OR 2D bottom barcode	4 mL	 13.50, 18, 77, 51, 71, 0.16
	OG00-675	.75 mL via sponge			1.5 mL	
	OG00-610	1 mL			2 mL	
	OH-600	.5 mL			2 mL	
With 2D bottom barcode	OG00-600.52P	2 mL	 Sponges for assisted collection (OG00-675 products only) 2D bottom barcode	 OR 2D bottom barcode	4 mL	 13.50, 18, 82, 51, 76, 0.16
	OG00-675.52P	.75 mL via sponge			1.5 mL	
	OG00-610.52P	1 mL			2 mL	
	OH-600.52P	.5 mL			2 mL	

† Weight is inclusive of all product configurations within referenced catalogue numbers.
 †† Total sample volume includes saliva and Oragene stabilization solution. May vary based on volume of saliva collected.
 © 2017 DNA Genotek Inc., a subsidiary of OraSure Technologies, Inc. All rights reserved. Patent (www.dnagenotek.com/legal/notices)



MAYDA: Manage Your Data

MAYDA - Manage your data

Tableau de bord de gestion de la vie privée

[Info](#)
[Médecin référent](#)
[Données de santé](#)
[Accès](#)
[Consentement](#)
[GDPR](#)
[Kit](#)
[Messagerie](#)
[Donation](#)
Romain Georges H A...

Consentement

Vous pouvez retrouver et télécharger ici le(s) consentement(s) que vous avez signé(s).
 Vous pouvez à tout moment retirer (ou reconfirmar) votre(vos) consentement(s) en le(s) désactivant(s) (ou en le(s) réactivant(s)) en utilisant le bouton curseur d'activation.

1. GEMS under_twelve ✓ [Télécharger ce consentement](#) Consentement actif

[Invitation à rejoindre d'autres études](#)

MAYDA - Manage your data

Tableau de bord de gestion de la vie privée

[Info](#)
[Médecin référent](#)
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[Accès](#)
[Consentement](#)
[GDPR](#)
[Kit](#)
[Messagerie](#)
[Donation](#)
Romain Georges H A...

Droits conférés par le GDPR

Via le portail de "Gestion de profil - Tableau de bord de gestion de la vie privée" vous êtes en mesure d'exercer tous les droits qui vous sont conférés par le GDPR.

Vous pouvez aussi accéder à la politique de vie privé en [cliquant ici](#).

Vous pouvez exercer:

- votre droit à l'information, notamment, via la page "Accès";
- vos droits à l'accès à la rectification via les pages "Info"; "Médecin référent", "Données de santé" et "Accès";
- vos droits de retrait du consentement, de l'effacement, de l'opposition au traitement et de la limitation du traitement via la page "Consentement";
- votre droit à la portabilité des données à caractère personnel en téléchargeant ici l'ensemble des données personnelles que nous avons collectées via le lien ci-dessous "Télécharger mes données".

[Télécharger mes données](#)

Pheno Boxes












PHENO Boxes

Via the different "boxes" below you can choose to communicate health data that are very important for the GEMS project: these data will help to "decipher" the alphabet of the genome and shed light on the interaction between genes.

You can communicate this information in stages and come back regularly to update it. Each completed box changes colour and appears in green.

Note that the last 'box' allows you to upload documents that you want to share.

 FBN1	 Information	 Family
 Ethnicity	 Aorta dimension	 Aorta medication
 Aorta surgery	 Aorta dissection	 Confounding factors

Aorta surgery

Romain Alderweireldt

Have you ever had surgery on the aorta (root and/or valve)?

Yes No

Can you tell us what type of aortic surgery you had?

Bentall

David

PEARS

Supra-coronair

I don't know

Can you tell us in millimeters what the largest measured diameter of your aorta was before surgery?

Largest diameter in millimeters

Have you had an aortic valve surgery?

Mechanics Bioprosthesis Plastic surgery (repair) No

Have you had mitral valve surgery?

Mechanics Bioprosthesis Plastic surgery (repair) No

Have you had surgery on the descending aorta ?

Aortic arch Endoprothèse Open surgery No

Go global!

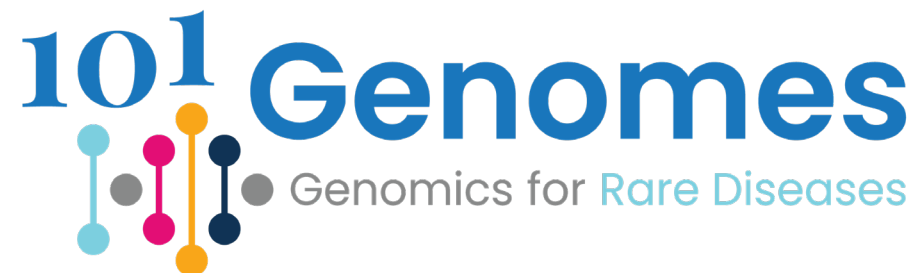
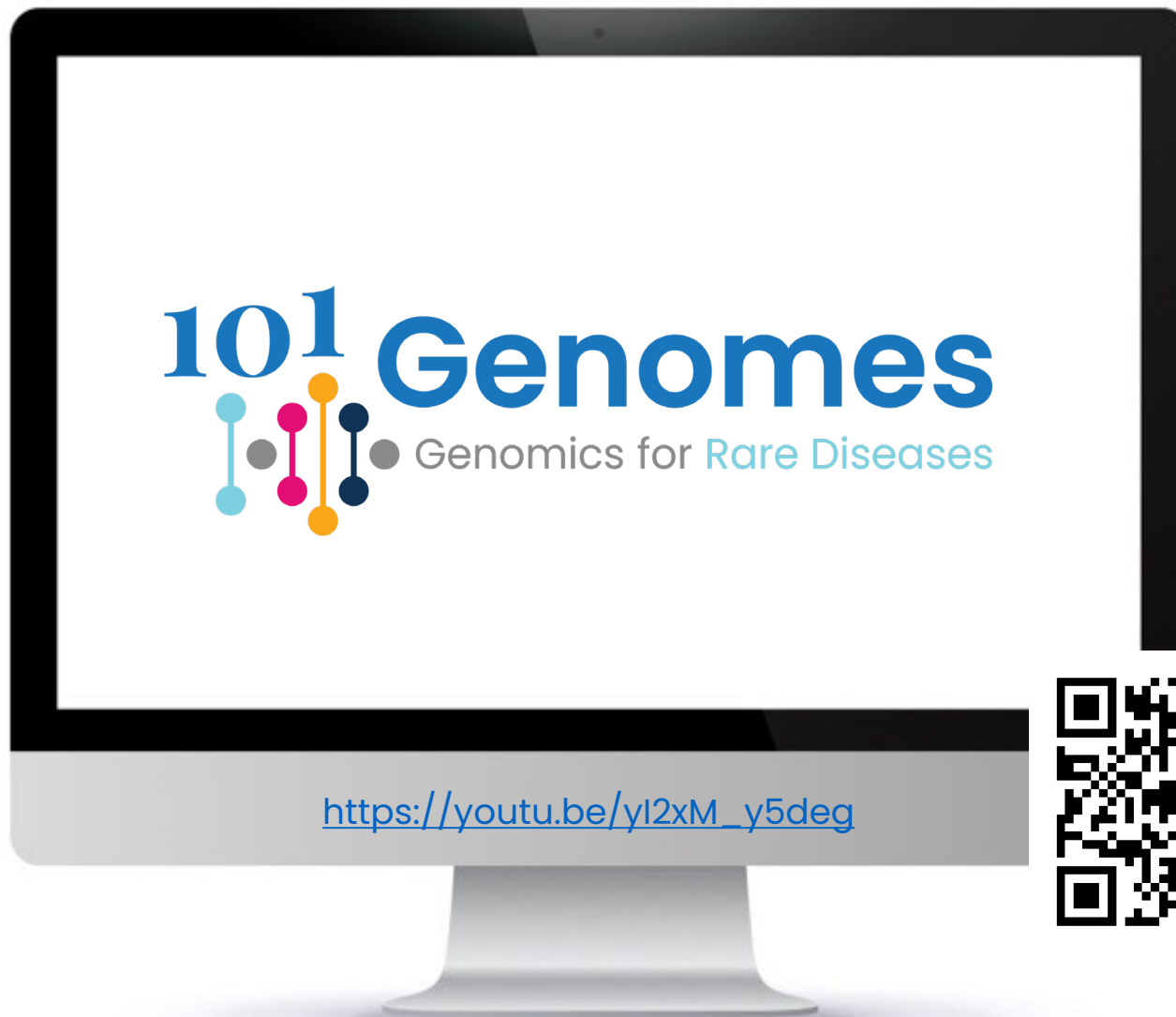
Join GEMS!



THE **MARFAN**
FOUNDATION



VASCERN



https://youtu.be/yI2xM_y5deg

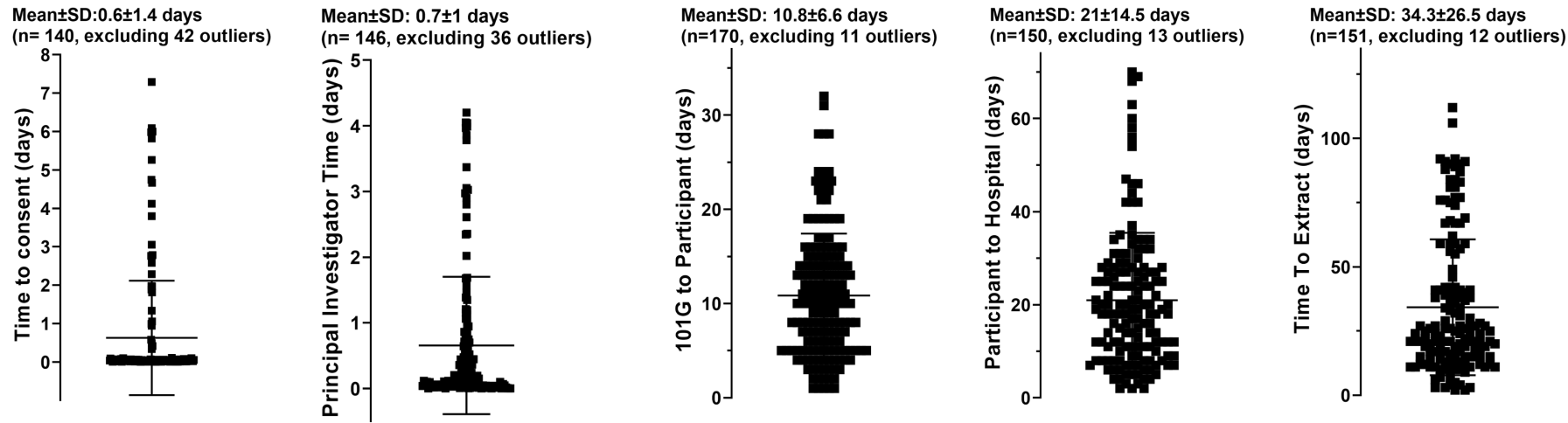
Join
GEMS!



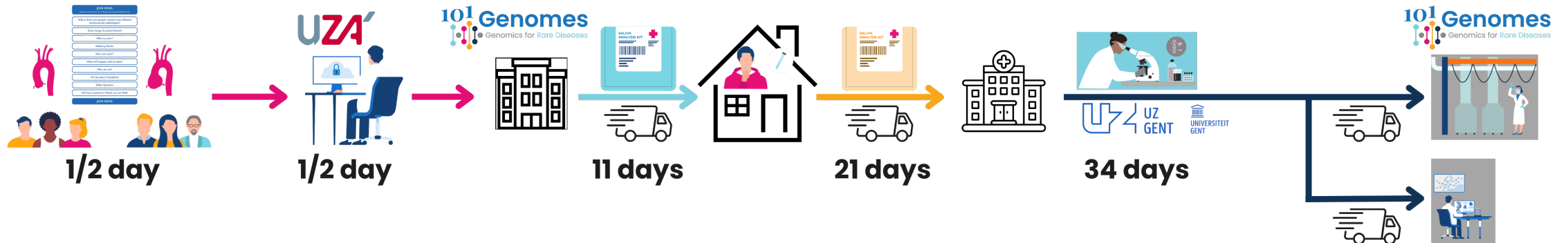
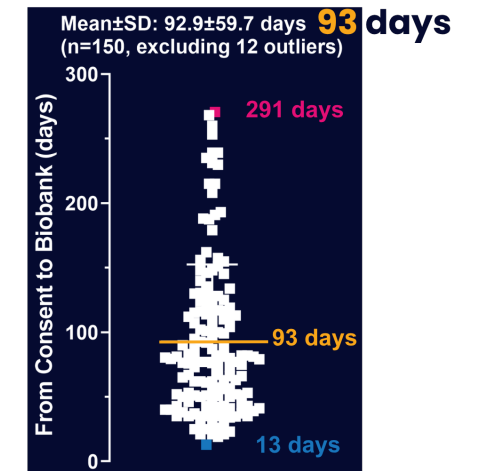
MAIN FINDINGS

December 2025: 182 consents, 93 days journey

Figure 1. GEMS-App accommodates all time frames



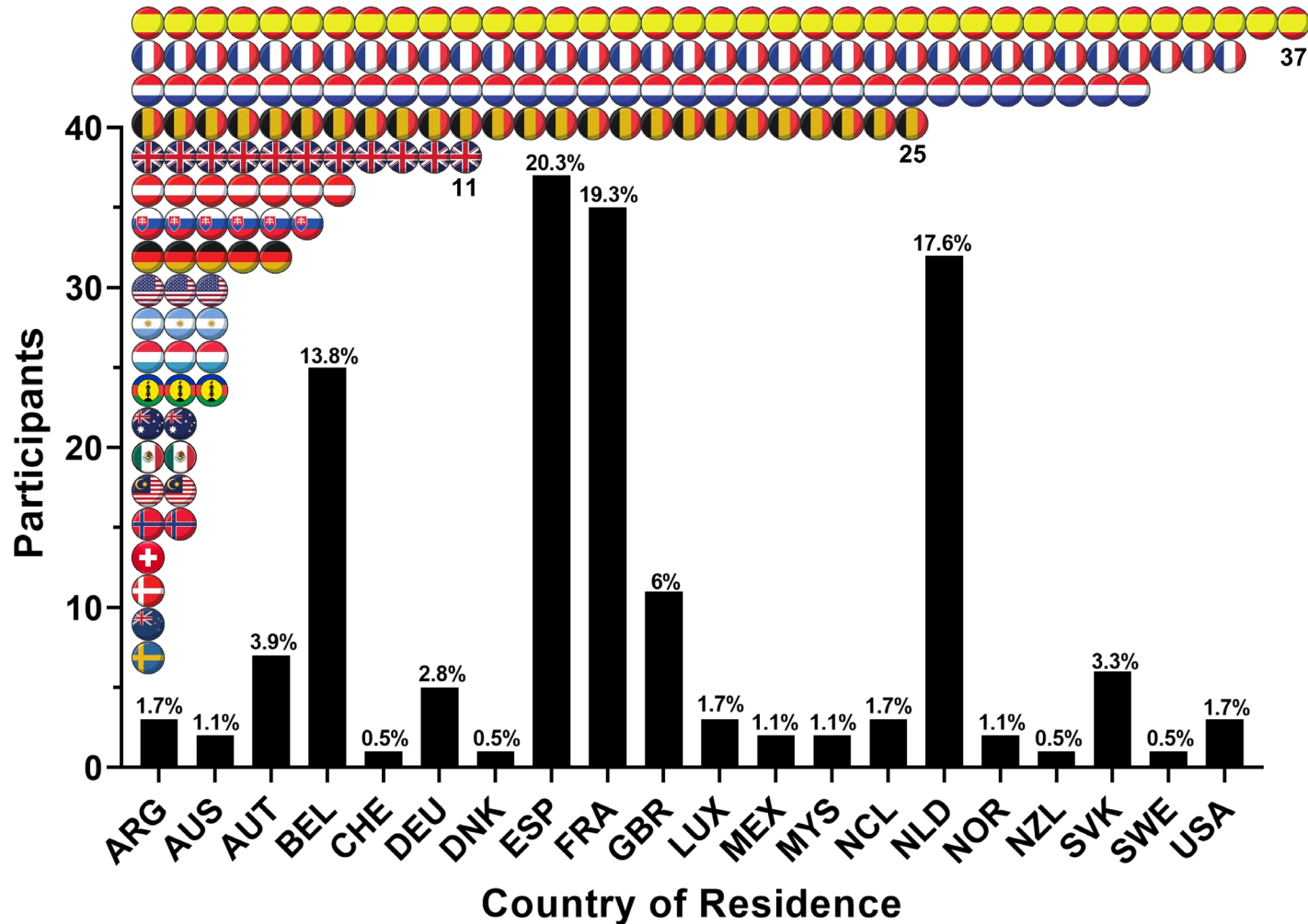
Complete GEMS Journey



20 Countries

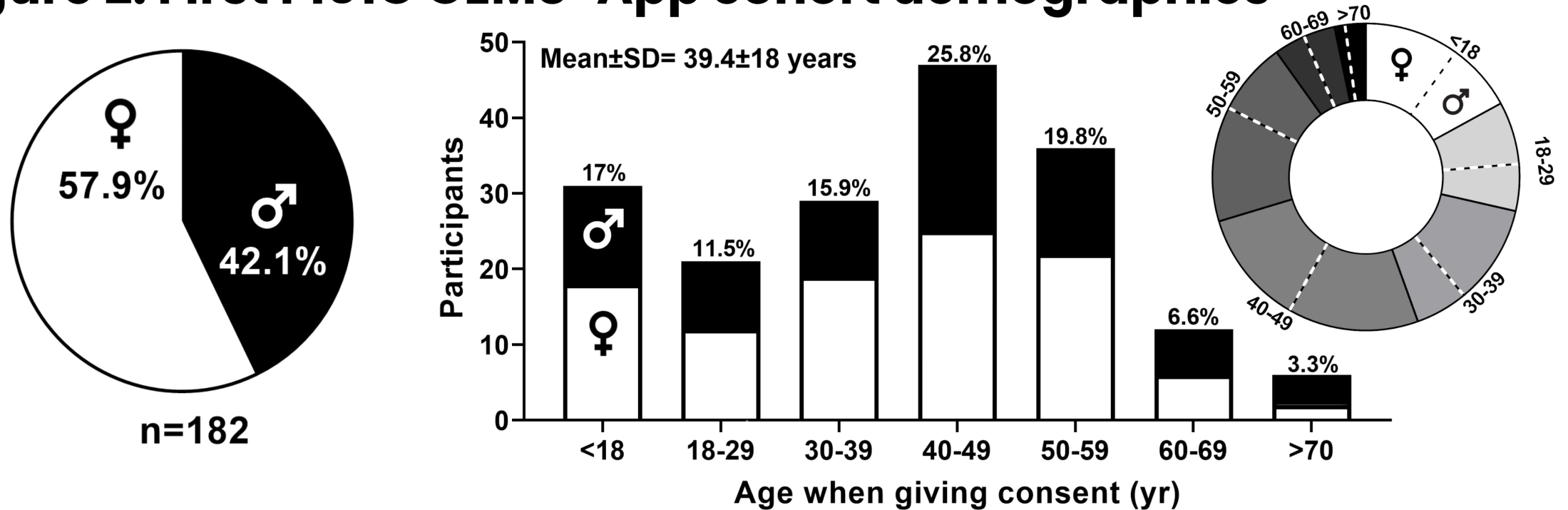


182 participants via Gems App from 20 countries: Belgium, France, Luxembourg, the Netherlands, Slovakia, Sweden, Norway, Austria, Spain, UK, Denmark, Swiss, USA, Australia, Mexico, Argentina, New Zealand, and Malaysia.



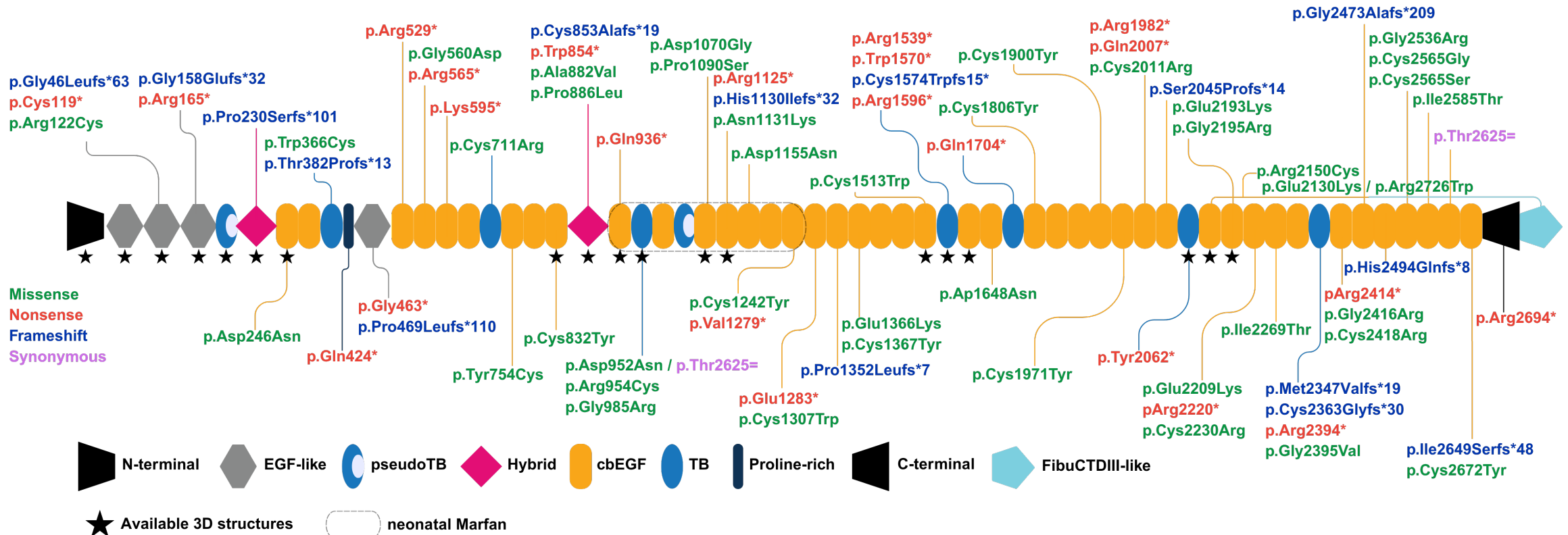
Demographics

Figure 2. First F101G GEMS-App cohort demographics

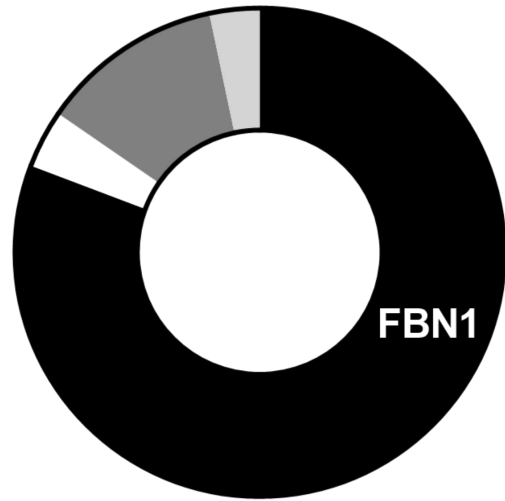


147 variants across *FBN1*

Figure 3. Variants across the entire Marfan causing *FBN1*

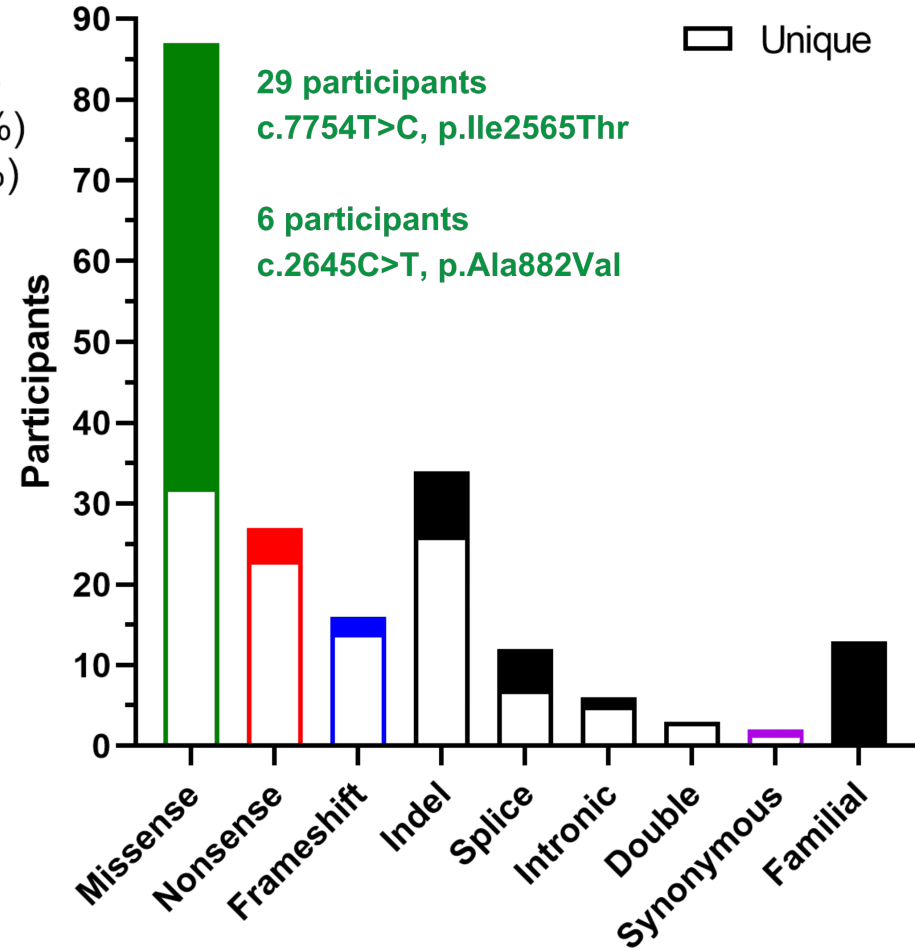


29 c.7754T>C (& 6 c.2645C>T)



Given Consents=182

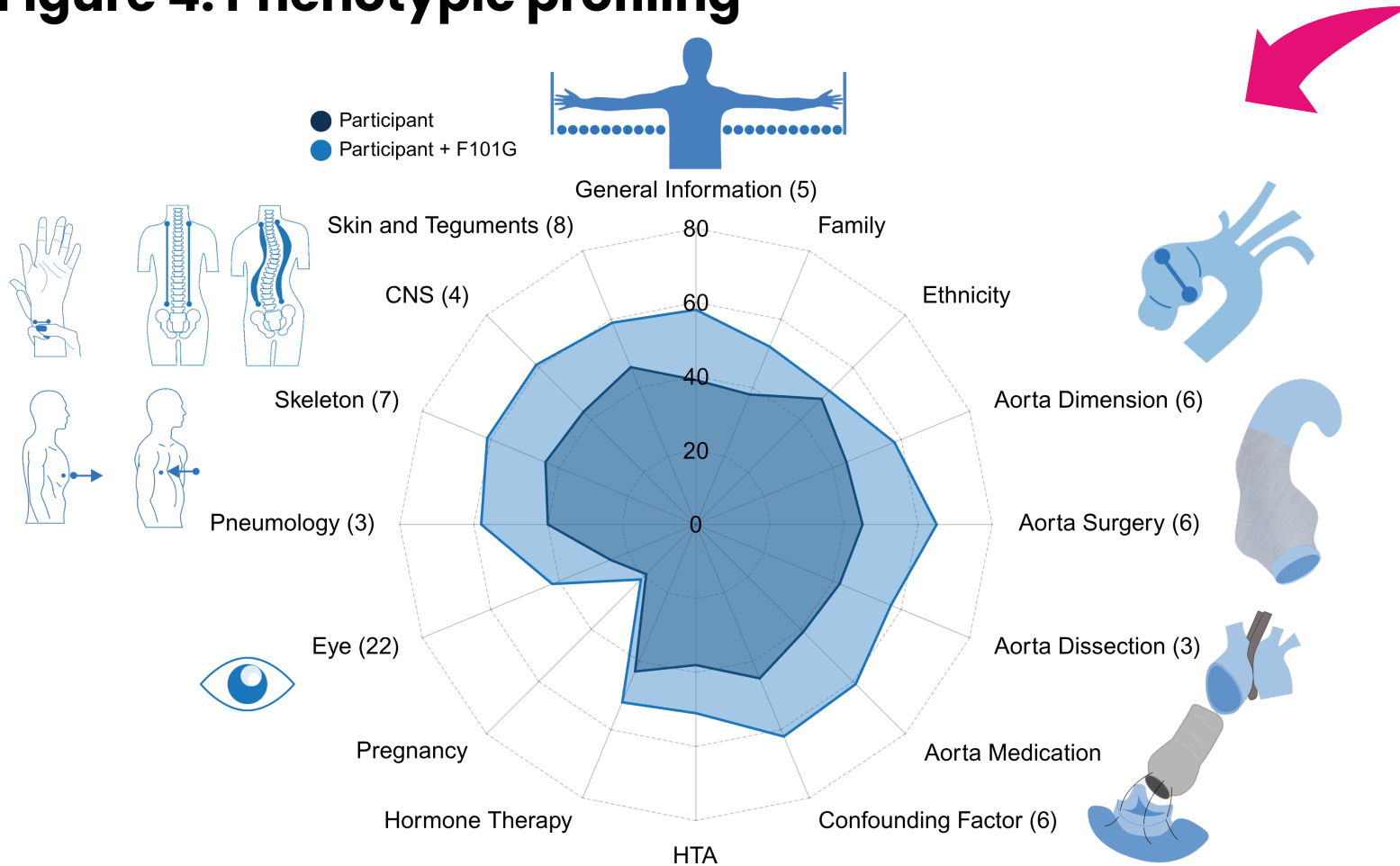
- FBN1 (147, 80.8%)
- Non FBN1 (7, 3.9%)
- Unknown (22, 12.1%)
- Unreported (6, 3.3%)



1/3 grey phenotypes & white data



Figure 4. Phenotypic profiling



DocuSign Envelope ID: 7ABA4EB8-8AAC-4412-BABF-EAB1CBC68FA9

DATA REPATRIATION AUTHORISATION (Version 02 of 14/03/2023)

I hereby authorize the 101 Genomes Foundation to collect the medical information from my medical file from the doctors who follow me and to repatriate all the computerized data from the medical images (CT-SCAN, MRI, etc.) to sequencing of my genome, exome and/or gene panel already performed in another context.

This authorization comes in the context of my participation in the GEMS research and my desire to participate in the 101 Genomes Foundation's initiative to nourish research for the greatest number of researchers.

Done at Brussels

First name: Romain Georges H

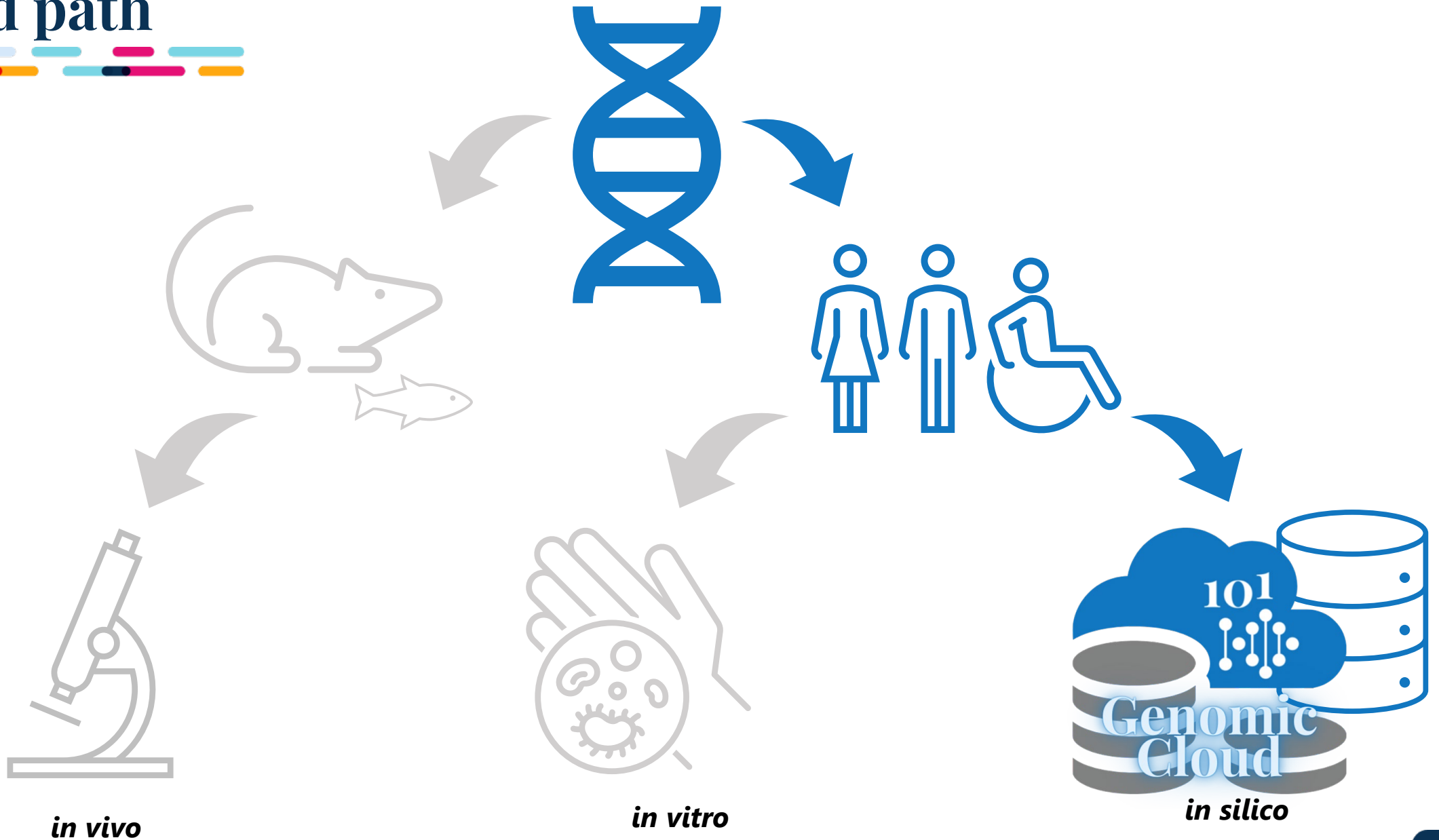
Name: Alderweireldt

Signature:

DocuSigned by:
Romain Georges H Alderweireldt
A077885D85B2E43428.....

CONCLUSIONS

A third path



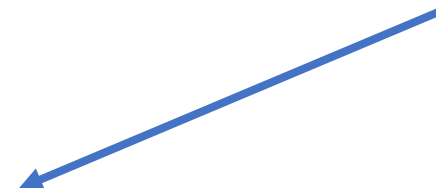
Digital data → Genomic Cloud



220
(38+182)
GENOMES



111
EXOMES



638
PANELS



220 WGS + 111 WES + 638 Panels = 969 NGS!!!



Data altruism organisation = DAO



THE **MARFAN**
FOUNDATION



Entrust



Query



DAO Recognition since 4 June 2025

16 juin 2025



SPF Economie, P.M.E., Classes moyennes et Energie
Rue du progrès 50 - 1210 Bruxelles

Ludvine Verboogen
Fondation 101 Génomes
N° BCE : 0684609172
Avenue de Sumatra, 6, 1180 Bruxelles

Notre référence : E3-DEE-2025-002260

Objet: Confirmation de la demande d'enregistrement dans le registre public national des organisations altruistes reconnues conformément à l'article XII.43 du Code de droit économique.

Madame Verboogen,

La demande faite par la Fondation 101 Génomes le 16 décembre 2024 conformément à l'article 6 de l'arrêté royal du 1^{er} octobre 2024 mettant en œuvre le règlement (UE) 2022/868 du Parlement et sur la gouvernance européenne des données et modifiant le régime du 1^{er} octobre 2024).

1. Evaluation

La demande d'enregistrement porte sur l'activité de la Fondation en matière de données conformément à l'article 2, 16) du règlement du Conseil du 30 mai 2022 portant sur la gouvernance européenne 2018/1724 (ci-après : règlement sur la gouvernance des données).

La Fondation 101 Génomes a, conformément à l'article 6 de l'arrêté, soumis les documents requis, à savoir :

- Le formulaire d'enregistrement des organisations altruistes
- Un extrait du casier judiciaire datant de moins de six mois
- 1^{er} du Code d'instruction criminelle, ou un document à caractère personnel qu'elle effectue dans le cadre des activités couvertes par cet enregistrement.

L'organe d'inscription a procédé à une vérification de la demande.

Il confirme que la demande d'enregistrement est complète et correcte.

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N° d'entreprise: 0314.595.348

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2. Décision

La Fondation 101 Génomes sera enregistrée dans le registre public national des organisations altruistes reconnues dans les meilleurs délais et peut utiliser le label « organisation altruiste en matière de données reconnue dans l'Union » dans ses communications écrites et orales, ainsi que le logo commun.

Cet enregistrement est valable à partir du 4 juin 2025 et est soumis aux conditions suivantes, conformément à l'article XII.44 du Code de droit économique :

- L'organisation altruiste en matière de données respecte, pendant toute la durée de ses activités, les conditions visées à l'article XII.43 du Code de droit économique ;
- Toute modification des informations fournies conformément à l'article XII.43 est communiquée par écrit ou par voie électronique à l'organe d'inscription dans un délai de quatorze jours à compter de la modification.

Conformément à l'article 19, paragraphe 5, alinéa 2, du règlement sur la gouvernance des données, l'organe d'inscription notifie votre enregistrement à la Commission européenne. En conséquence, la Fondation 101 Génomes sera inscrite dans le registre public de l'Union des organisations altruistes en matière de données reconnues.

L'enregistrement en tant qu'organisation altruiste de données au titre du Règlement sur la gouvernance des données n'implique pas un contrôle du respect du Règlement général sur la protection des données (RGPD). Il appartient à l'organisation altruiste de données de s'assurer du respect du RGPD pour les traitements de données à caractère personnel qu'elle effectue dans le cadre des activités couvertes par cet enregistrement.

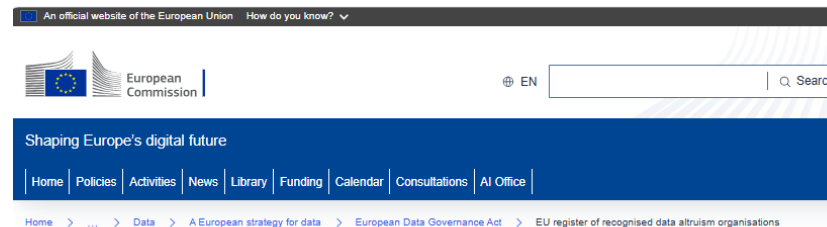
3. Informations relatives au recours contre la présente décision

Un recours en annulation peut être introduit contre la présente décision auprès du Conseil d'État. La demande d'annulation doit être envoyée par voie postale en recommandé au Conseil d'État, rue de la Science 33, 1040 Bruxelles, ou par voie électronique (<http://www.raadvst-consetat.be/?page=e-procedure&lang=fr>), dans un délai de 60 jours à compter de la présente notification.

Fait à Bruxelles, le 16 juin 2025.

Pour l'organe d'inscription,

Jérôme Debrulle
Directeur général
Direction générale Réglementation économique



EU register of recognised data altruism organisations

This EU register of recognised data altruism organisations has been established in the framework of the Data Governance Act (DGA), a key pillar of the European strategy for data.

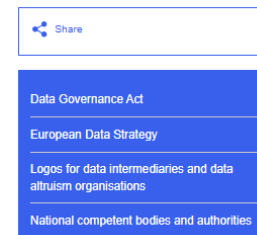
The DGA creates the right conditions for individuals and companies that when they share their data voluntarily for the benefit of society, it will be handled by trusted organisations based on EU values and principles. These organisations that make relevant data available based on data altruism will be able to register as 'data altruism organisations recognised in the Union'.

These entities must have a not-for-profit character and meet transparency requirements as well as offer specific safeguards to protect the rights and interests of citizens and companies who share their data. In addition, they must comply with the rulebook which will lay down information requirements, technical and security requirements, communication roadmaps and recommendations on interoperability standards. The rulebook will be developed by the Commission, in close cooperation with data altruism organisations and other relevant stakeholders.

Individuals and companies now have the tools to give their consent or permission to make their generated data available – voluntarily and without reward – to be used for objectives of general interest. Such data has enormous potential to advance research and development of services, including in the fields of health, environment, and mobility.

This page maintains a public register of all recognised data altruism organisations in the European Union.

Member States will notify the Commission as and when data altruism organisations are registered.



Recognised data altruism organisations notified by Member States

The list below indicates the countries where the data altruism organisations are registered:

- Spain: [Associació Dades pel Benestar Planetari - Datalog](#)
- Belgium: [101 Genomes](#), [European Brain Data Hub \(EBDH\)](#)

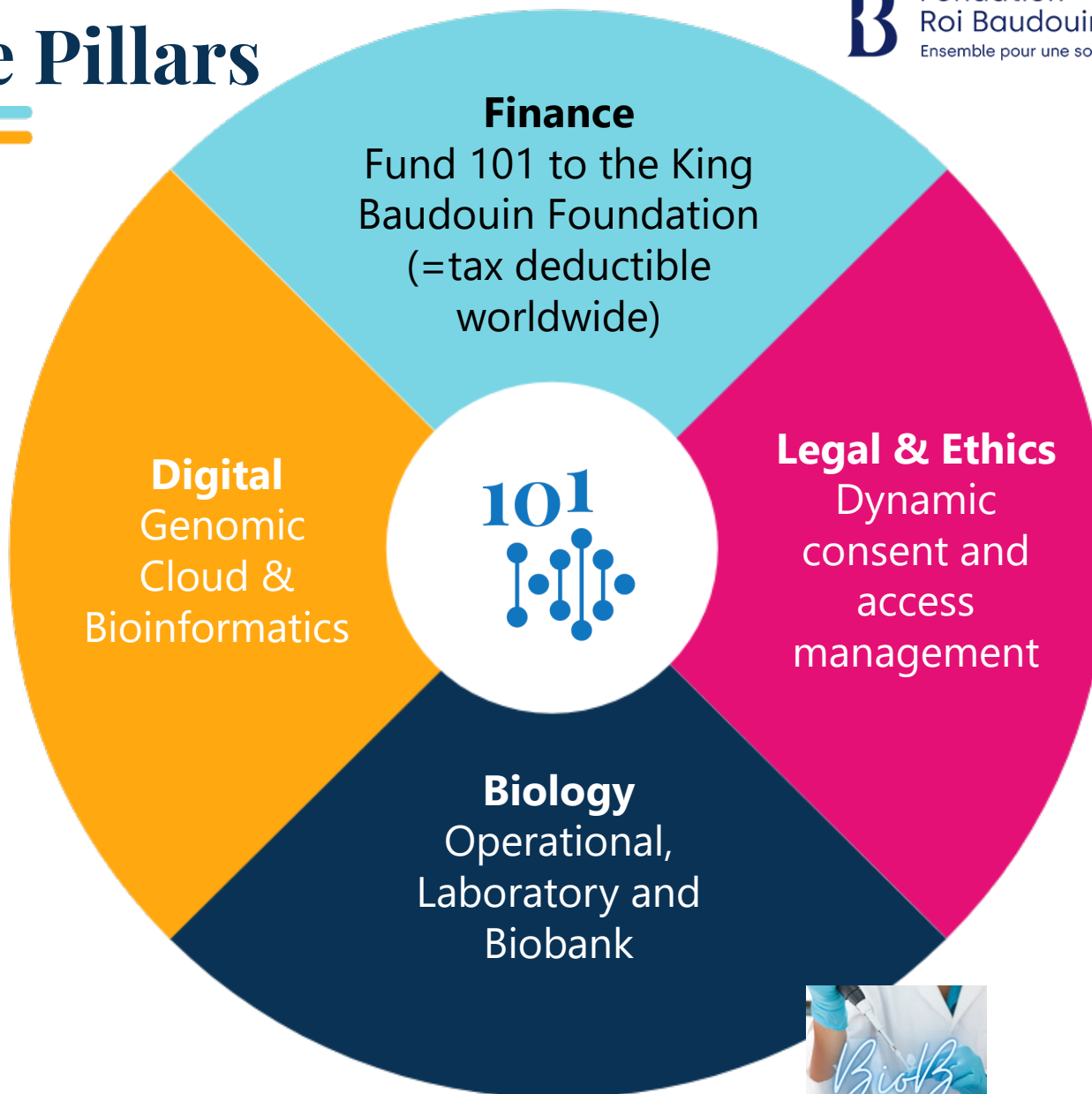
The map shows in which countries the data altruism organisations provide their services:



<https://digital-strategy.ec.europa.eu/en/policies/data-altruism-organisations>



Four re-usable Pillars



MAYDA
Manage your data
GEMSAPP



101 WGS MFS target achieved!
Exponential model
Ready to be extended to other genomics projects

101 Loey-Dietz Project: GELDS



Researchers dedicated ...



Prof Bart LOEYS



Prof Marianna PARLATO

... specific funding...

Fund Plisnier - Fund Tournay-Dubuisson
 managed by the King Baudouin Foundation

CANDIDATES APPLICATION FORM

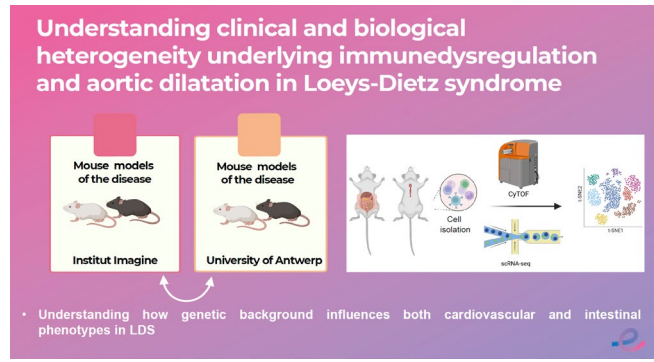
1. Project information

Title of the project

GELDS: Unlocking Genomic Modifiers to Transform Care in Loey-Dietz Syndrome.

Scientific abstract

Rare diseases collectively affect millions of individuals worldwide, yet progress in their diagnosis and treatment remains disproportionately slow. Two key barriers hinder advancement: the chronic shortage of high-quality data, which is compounded by limited patient involvement, and the continued reliance on traditional laboratory-based approaches that insufficiently integrate modern genomic and computational innovations. As a result, valuable opportunities offered by genomics, transcriptomics, bioinformatics, and integrative data science are often missed. To overcome these limitations, the *Genomic Modifiers for Loey-Dietz Syndrome (GELDS)* project proposes a disruptive research model, designed to transform the way data on rare diseases is collected, processed, and exchanged.



...a patient project leader



Aliénor LECOMTE
alienor.lecomte@f101g.org

One Genome at a time...

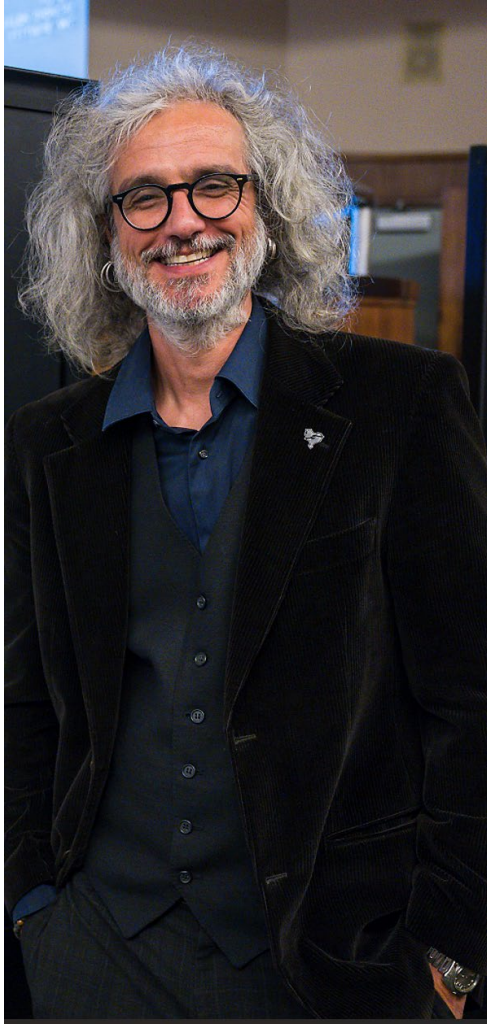


**Building a
Citizen-
Centred
Genomic
Infrastructure**



**...one rare
disease at a
time.**

Thank you!



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VERSBRAEGEN, Félix CARATI, Loris GIORDANO, Aliénor LECOMTE, Guillaume SMITS
& Aurélien ALDERWEIRELDT