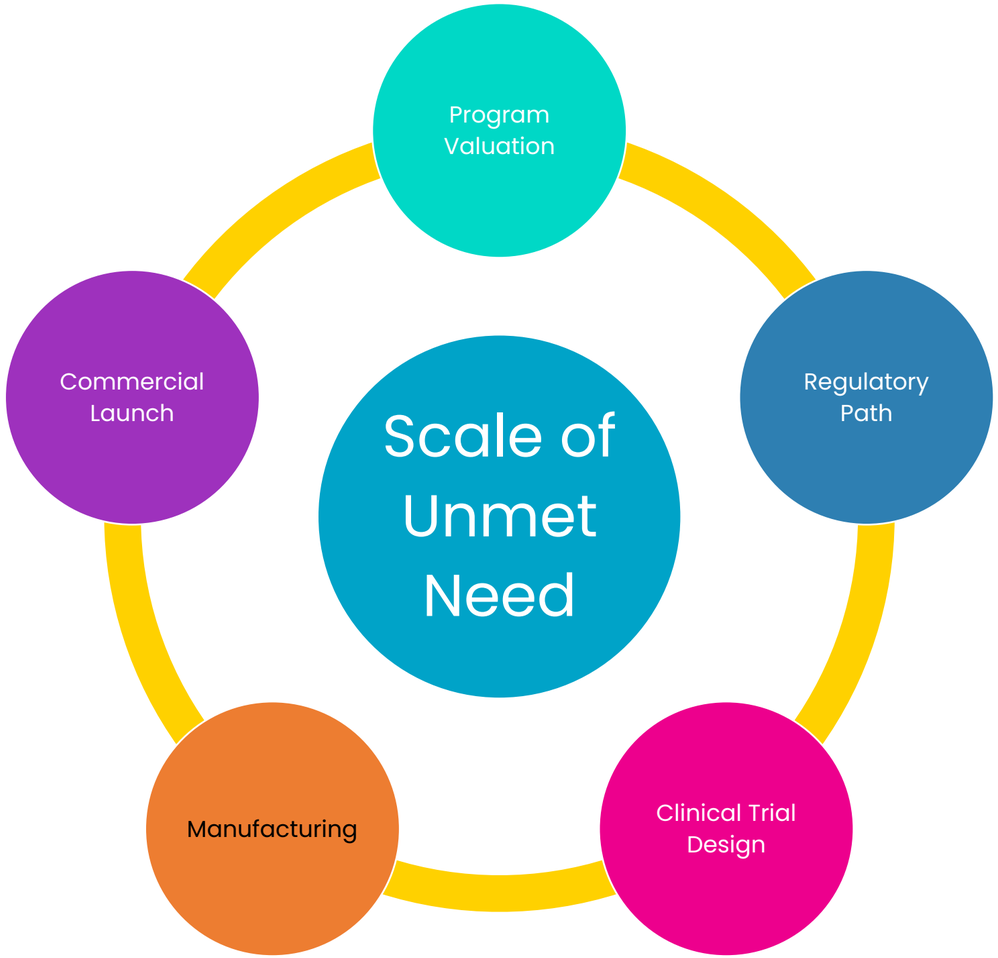
GeneScape

Your Compass for Navigating
Rare, Ultra-Rare and
Orphan Disease Epidemiology

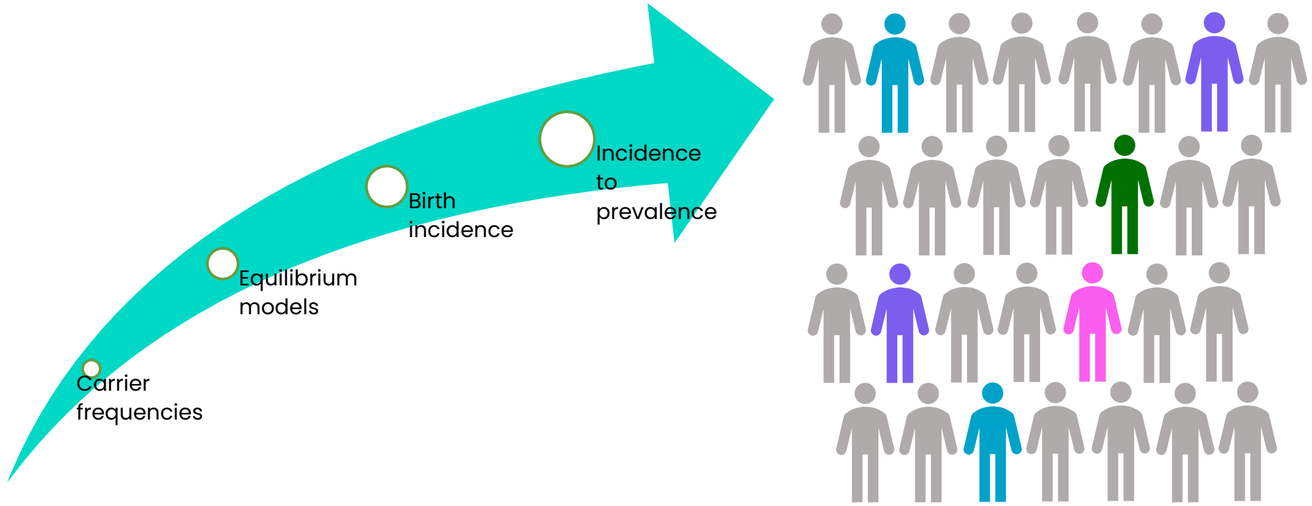
A Major Challenge in Orphan Drug Development:

Understanding the scale of unmet need is central to drug development strategies



GeneScape's Unique Approach:

Population genetics-based modeling



Major Impacts to Development Strategy

- Independent of diagnosis
  - can reveal hidden or overlooked patient subsets
- Improved allocation of resources
  - clinical trials & manufacturing
- Support regulatory filings

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How Does GeneScape Compare?

Best estimate of total number of CF patients within the US (c.2017):

28,675

95% Confidence Range: 25,982 – 30, 698

Estimated number of children born in the US with CF in 2017: **1,081**

95% Confidence Range: 995 – 1,175

CYSTIC FIBROSIS FOUNDATION
ADDING TOMORROWS

Total number of people with CF in the US registry in 2017†:

29,887

Newly diagnosed cases in 2017: **880**

Median age at diagnosis: 3 months

†:CF Foundation Annual Report 2017

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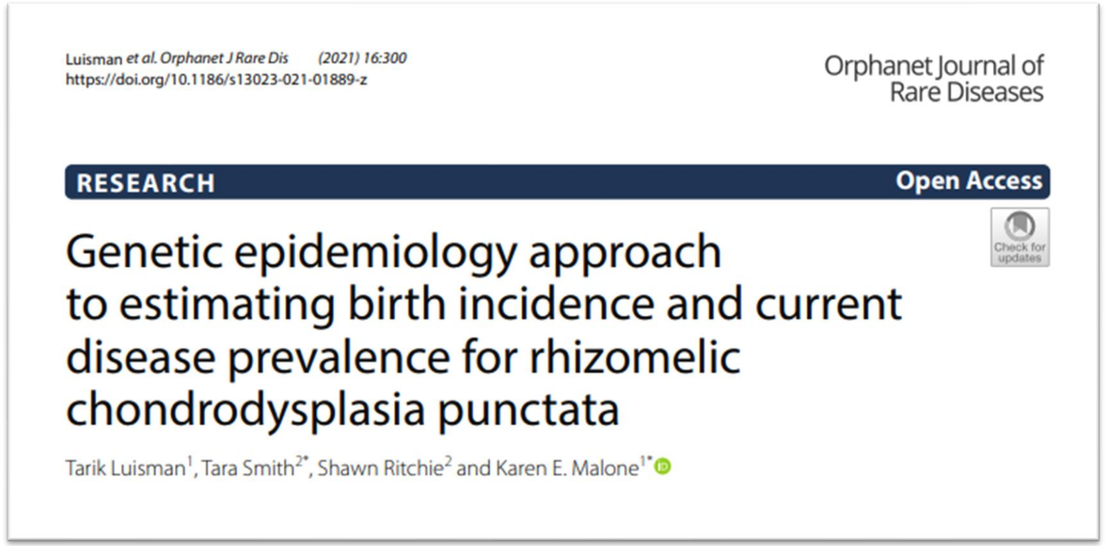
GeneScape's Collaborations and Partnerships



Luisman *et al. Orphanet J Rare Dis* (2021) 16:300
https://doi.org/10.1186/s13023-021-01889-z

Orphanet Journal of Rare Diseases

RESEARCH Open Access

Genetic epidemiology approach to estimating birth incidence and current disease prevalence for rhizomelic chondrodysplasia punctata

Tarik Luisman[1], Tara Smith[2*], Shawn Ritchie[2] and Karen E. Malone[1*]

OPUS GENETICS®

Pharming®

ASTHERNA

ProQR®

MLD MED-LIFE DISCOVERIES LP

McGill

GFPD GLOBAL FOUNDATION for PEROXISOMAL DISORDERS

FOXG1® RESEARCH FOUNDATION

University at Buffalo Department of Biological Sciences

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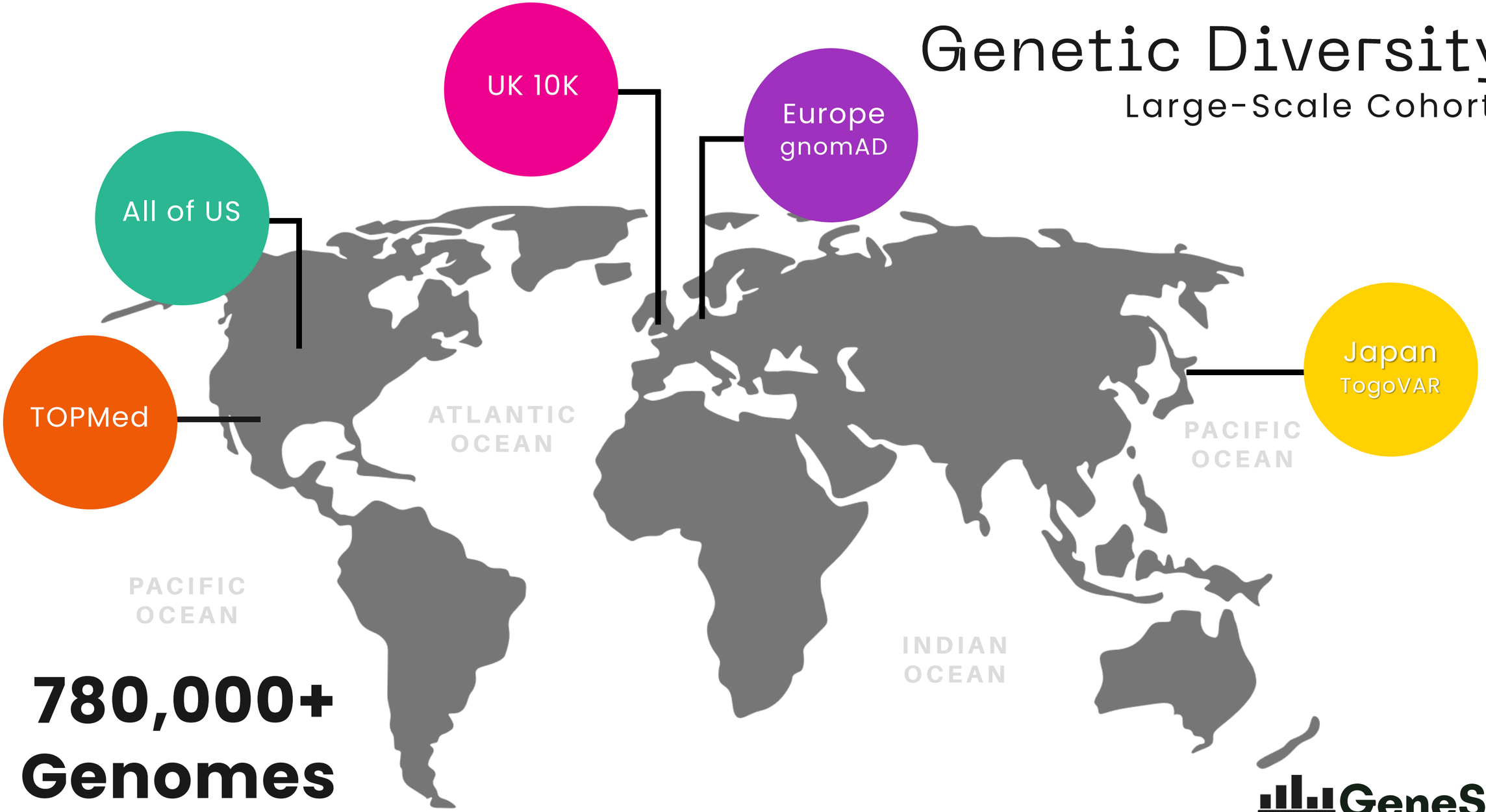
Genetic Diversity

Large-Scale Cohorts



780,000+ Genomes

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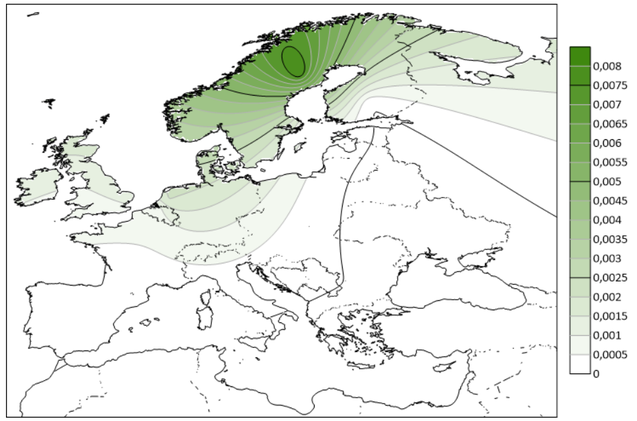
Increased resolution of variant geography

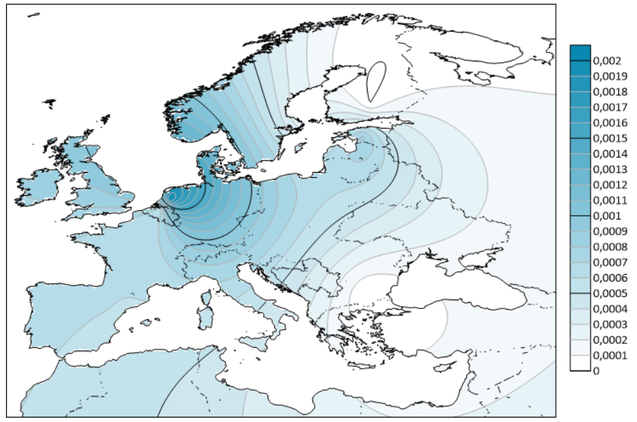
PEX1-driven Zellweger Spectrum Disease

- Highly variable disease with impact to life expectancy, depending on the genotype
- Two main variants (mutations) drive these populations in Europe
- These data can be used to determine the regions with highest probability of patients.

PEX1, p.I700fs



PEX1, p.G843D



C. Argyriou et. al., in preparation

Meet Our Team

Karen Malone, PhD
CEO

Tarik Luisman
Director Bioinformatics &
Digital Systems

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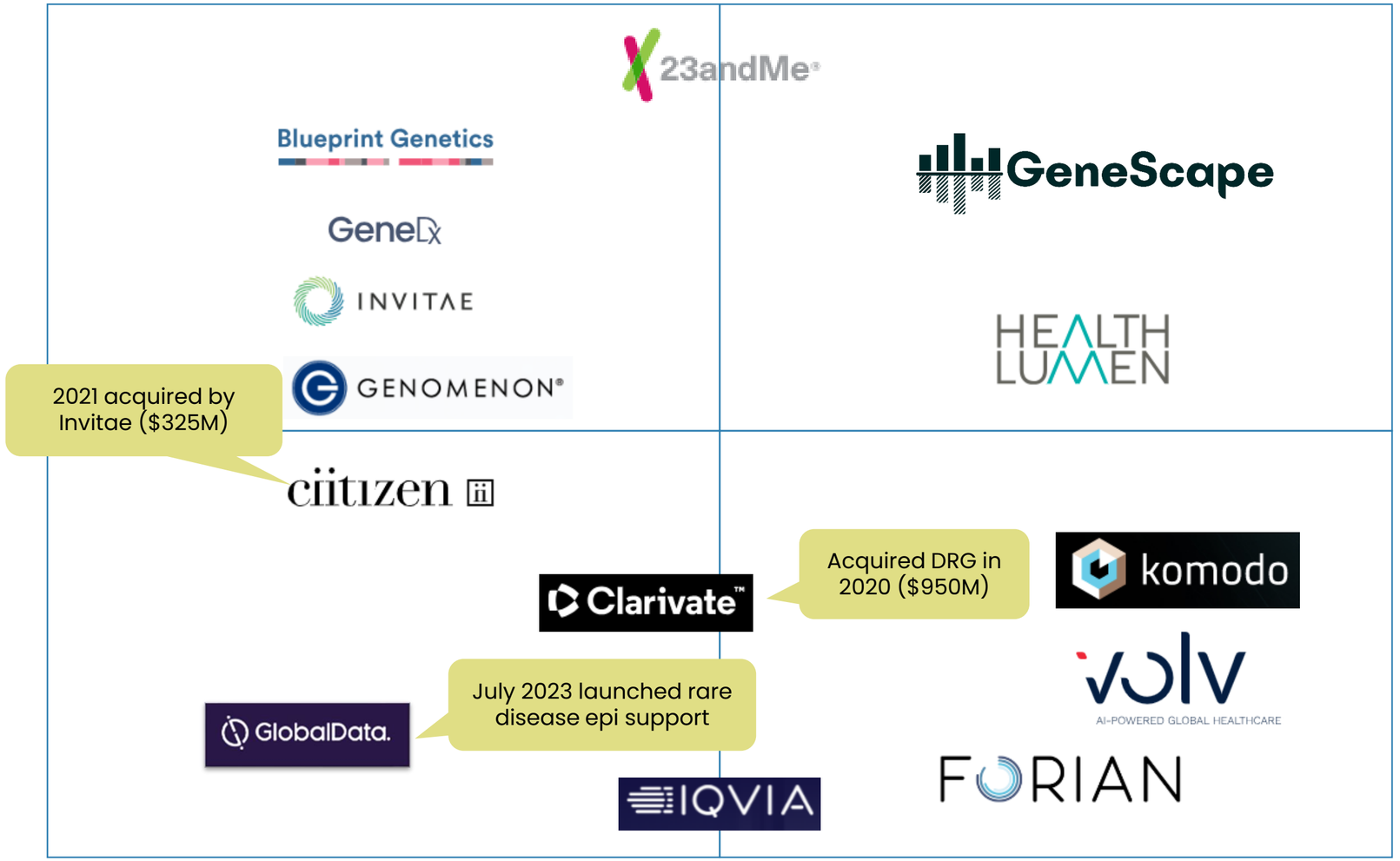
Communications & Strategy

Mathijs Gaastra
Data Analyst

GeneScape

Our Unique Positioning



Where to next?

Access to Capital

Continue Bootstrapping	Exploring non-dilutive strategies	Open to Seed Investment

Future Milestones

- Expect to top 1M+ genomes in H1/2024
- Add 2-4 FTE in the coming year
- Migrate to high performance computing environment
- Monetize MVP for literature-based tool in 2024

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