

Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.

Pedigree Questionnaire Results

Clinical and Phenotypic Data Capture Work Stream

Overview

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27 Responses

9 Driver Projects

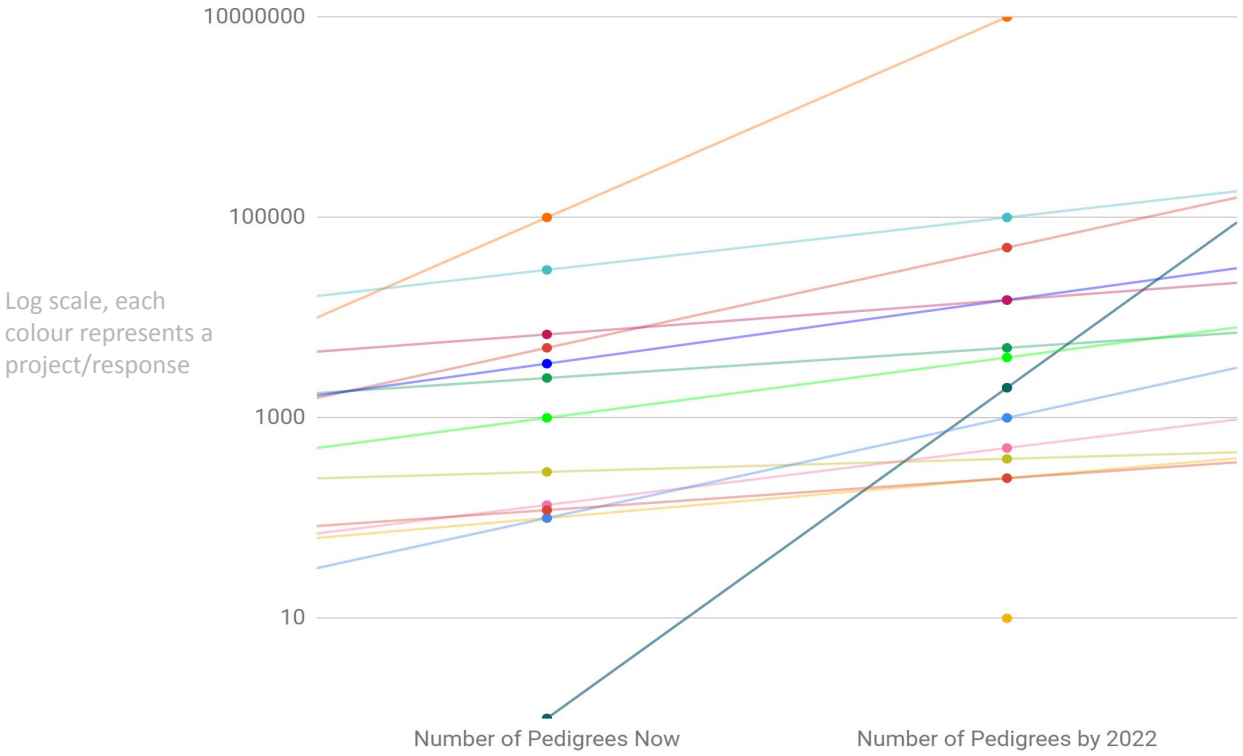
- Monarch Initiative
- GEL
- AGHA
- NCI GDC + NCI DCF
- EGA
- EJP-RD
- H3Africa
- SPHN
- BRCA Challenge

\+ Responses from other resources/databases such as Kids First Data Resource

20 responses **(74%)** had indicated they use or plan to use pedigree data

Pedigrees Now and in the Future

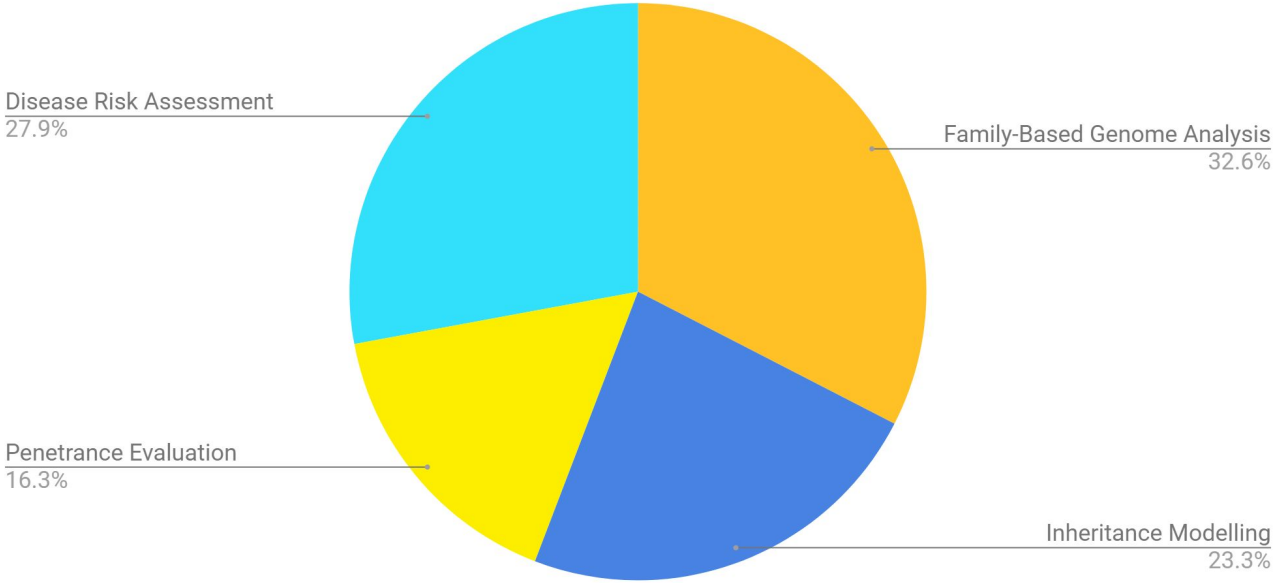
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Almost all responses indicated that they expect to see an increase in the number of pedigrees collected/used in the next few years

What is pedigree data used for?

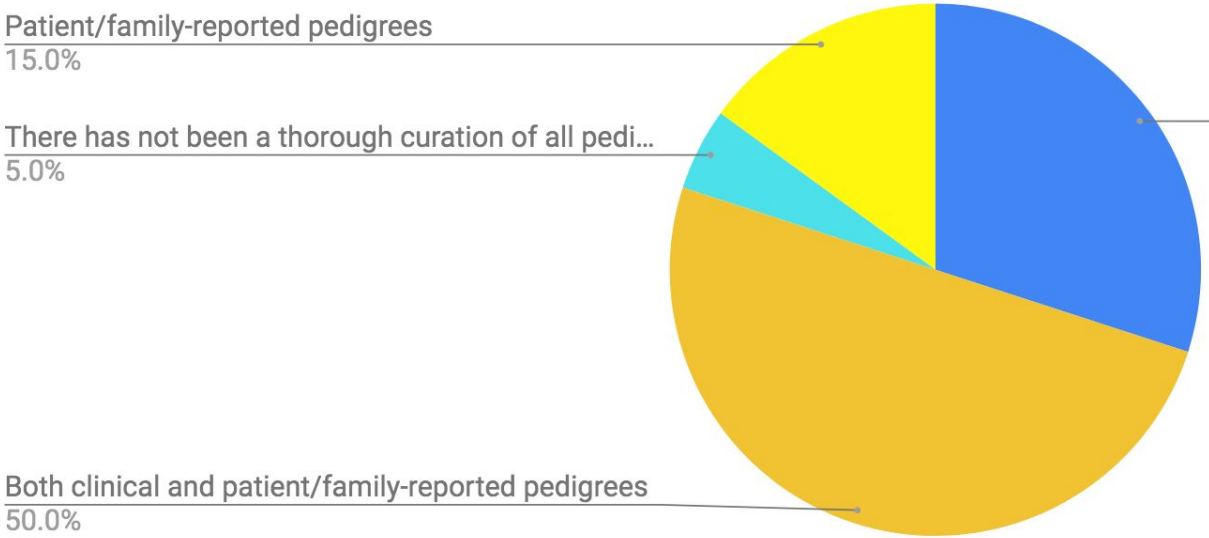
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In addition, over **half** of responses indicated that family-based genome analysis is the *most important use* of pedigree information

How is pedigree information collected?

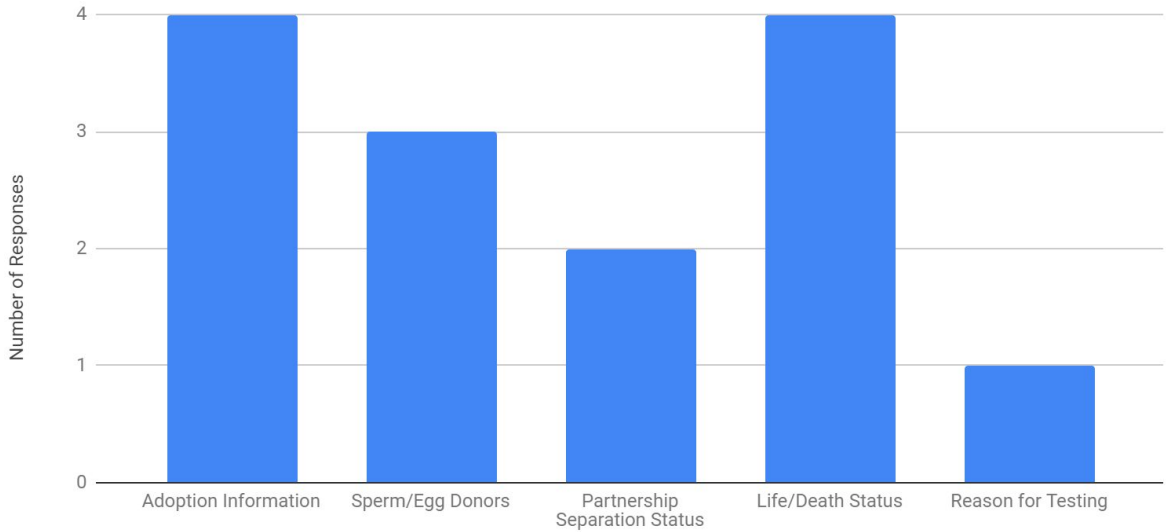
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The majority of responses collect information from **both** clinically and from patient/family reported pedigrees

Is there information you wished you were collecting, but aren't?

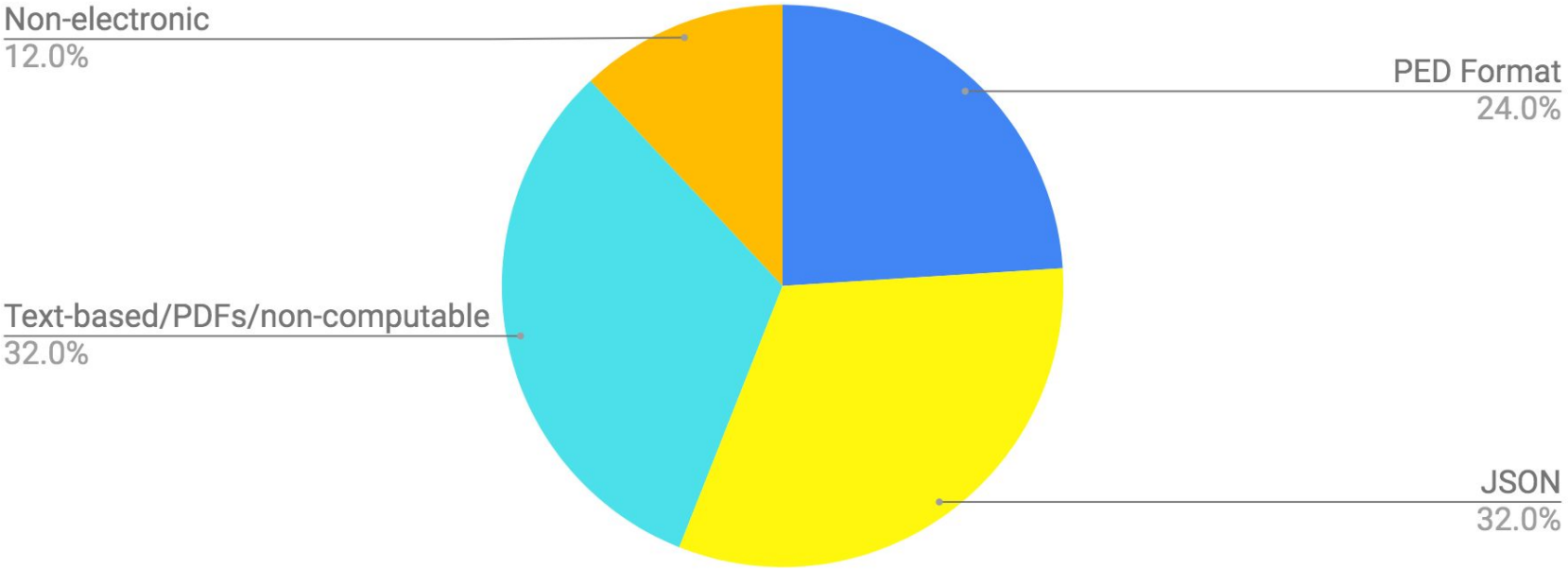
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Several responses indicated that there is information that they are *not* currently collecting, that would be useful to have

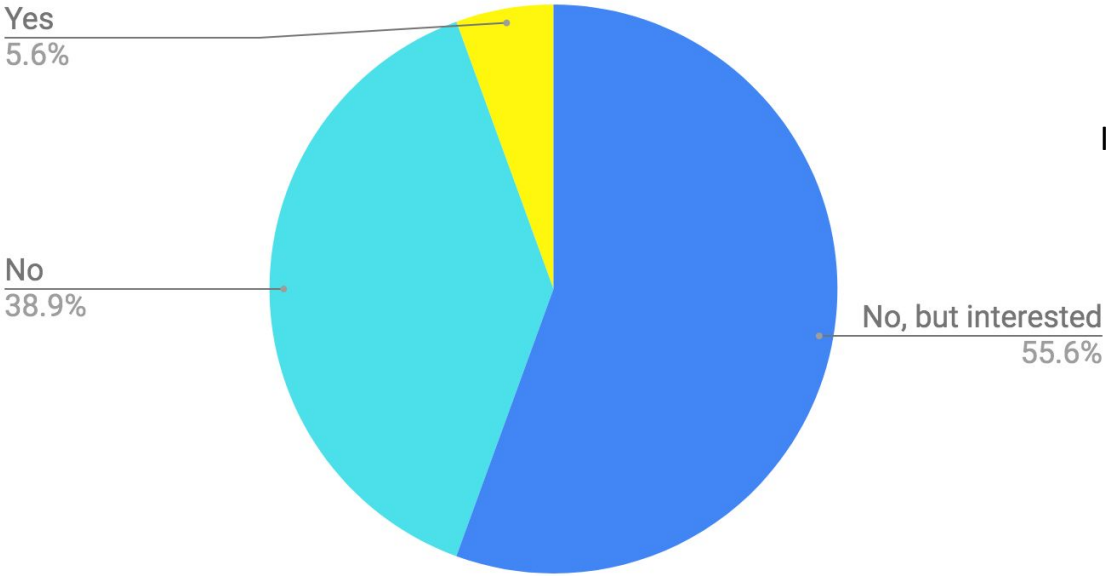
In what formats do you currently receive pedigree data?

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The majority of responses are receiving pedigree data in a **JSON format** or a **text-based/non-computable** format (ie. PDFs)

Do you recieve pedigree data directly from the EMR/EHR system?



The majority of responses **do not** receive data from the EMR/EHR system, but many are interested in doing so

In some projects, data is also received from the following 3rd party tools:

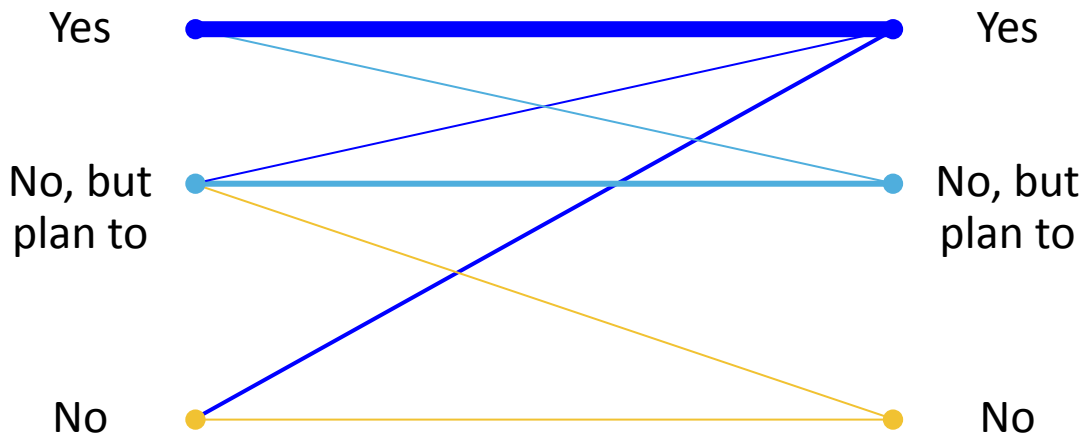
- PhenoTips
- Invitae Family History Tool
- ItRunsInMyFamily
- Cyrillic
- Ancestry health
- Trakgene
- CancerIQ
- Progeny

Data Import/Export

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Do you import/receive data from other tools and services?

Do you export data to other tools and services?



Note: Line thickness corresponds to number of responses

Most responses *currently*, or *plan to*, import/export data to other tools and services

Planned Export Tools/Formats

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To which tools/services do you (plan to) export pedigree data?

- GDC or GEN3 API
- Publications
- Paint
- Redcap Database
- FHIR
- Aligent Alissa
- In-house databases
- Genotype refinement pipelines
- Portal visualizations

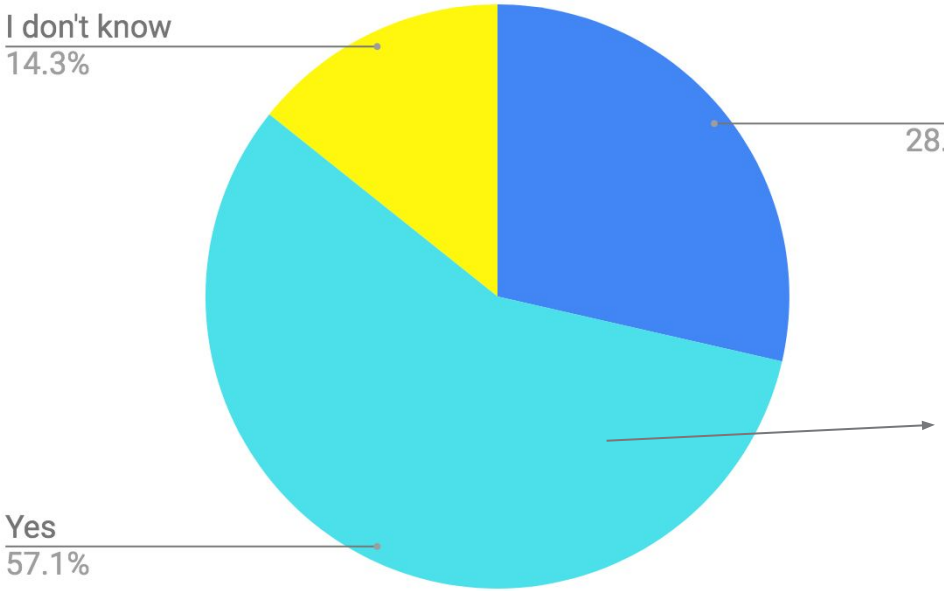
In what formats do you (plan to) export pedigree data?

- PED
- PFB
- PhenoTips JSON, GEL JSON, Custom JSON
- HL7 FHIR
- Powerpoint
- PDF of manual record
- GEDCOM
- BOADICEA
- Mendel

Key Takeaway: There are no common tools/services or formats used with pedigree data export

Are you experiencing challenges with standardization?

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Examples include:

- Hand drawn pedigrees
- Lack of computability, PDFs
- Representation of complex families
- Inability to share data easily
- Lack of integration with analysis tools
- Difficulty defining relationships and tree properties

The majority of responses (57.1%) are having pedigree standardization challenges

Would a standard have helped?

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2/3 of respondents said

YES

a standard would have helped in some way

(No respondents said ‘NO’!)

Conclusion

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Almost all responses indicated that they would be an **active** or **passive** participant in the development of a pedigree data standard.

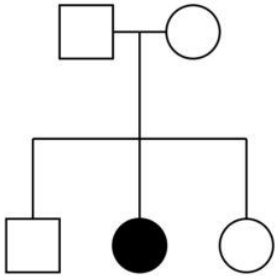
Many thanks for your participation in the pedigree questionnaire!

Please contact Lindsay Smith (lindsay.smith@ga4gh.org) if you have any questions.

Clinical & Phenotypic Data Capture Work Stream

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Supports the clinical adoption of genomics through establishing standard ontologies and information models to describe the clinical phenotype for use in genomic medicine and research, including the capture and exchange of information across electronic health records, research systems, registries, journals, and patient platforms



How can we help those working with pedigree data?

- Actively working on the development of a new pedigree standard that meets the needs of the community
- Presentations/seminars on formats, tools, and related topics of interest
- Contributors come from a wide variety of projects and organizations
- Pilot implementation support
- Bi-weekly meetings on Wednesdays at *12pm PT/3pm ET/ 8pm BST/9pm CEST/5am (next day) AEST, etc.* (19:00 UTC). All are welcome!

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