

PATIENTS - PURPOSE - PROGRESS



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RESULTS

Current research platform capabilities include filtering for real-

genomic instability HRD scores associated with specific

Affiliations of access-requesting users included academic

The top three themes cited by users for accessing the

institutions (35%, N=30), community hospitals (57%, N=49),

database included clinical research, patient care, and genetic

57%

Figure 2. Affliations of Access-Requesting Users and Themes

Genetic Testing

and other (e.g., laboratory, pharmaceutical, private clinician,

germline gene variants).

etc., 8%, N=7) (Figure 2).

Cited by Users for Accessing Database

Academic Institutions (N=30)

Community Hospitals (N=49)

Other (N=7)

time cohort comparisons across clinical variables (e.g., viewing

OBJECTIVES

- Studies that examine the real-world clinical impact of germline hereditary cancer, tumor mutational landscape, and homologous recombination deficiency (HRD) in medical decision-making and therapy are lacking.
- This is partly due to challenges in compiling structured data from multiple sources outside of clinical trials.
- To address this need, we created a datasharing platform from reliable patient datasets that combines germline and tumor genetic test results, family history, management, and
- Here, we describe the structure and composition of this registry and initial user interest.

- All patients identified as female, the median age of cancer diagnosis was 63, and the majority of patients had ovarian cancer (Table 1).
- Most patients had a positive family history of cancer (78%, N=3,507), with a median of 2 reported familial cases.
- A total of 2,877 patients reported and classified germline or tumor variants (deleterious, suspected deleterious, variants of uncertain significance) were observed.

A total of 4,521 patients were included in the registry at version 1 launch.

Table 1. Patient Characteristics (Total Registry Cases N=4,521)

Self Reported Ancestry, n (%)		Cancer Type ^b , n	
Ashkenazi Jewish	34 (0.8%)	Ovarian, Fallopian, Peritoneal	5,088
Asian	138 (3.1%)	Endometrial/Uterine	144
Black/African	318 (7.0%)	Other	721
Hispanic/Latino	310 (6.9%)	Detected Germline Variants, n	2,245
Middle Eastern	18 (0.4%)	Germline Status per Patient ^c , n	
Multiple Ancestry	274 (6.1%)	Deleterious	301
Native American	25 (0.6%)	Suspected Deleterious	65
Other/Unspecified	531 (11.7%)	VUS	1,217
Pacific Islander	2 (0.04%)	No Variant	2,937
White/Non-Hispanic	2,871 (63.5%)	Detected Tumor Variants, n	632
Positive Family Cancer History, n (%)	3,507 (77.6%)	Tumor Variant Classification ^b , n	
HRD Status ^a , n (%)		Deleterious	393
Positive	1,342 (29.7%)	Suspected Deleterious	25
Negative	2,751 (60.8%)	VUS	214

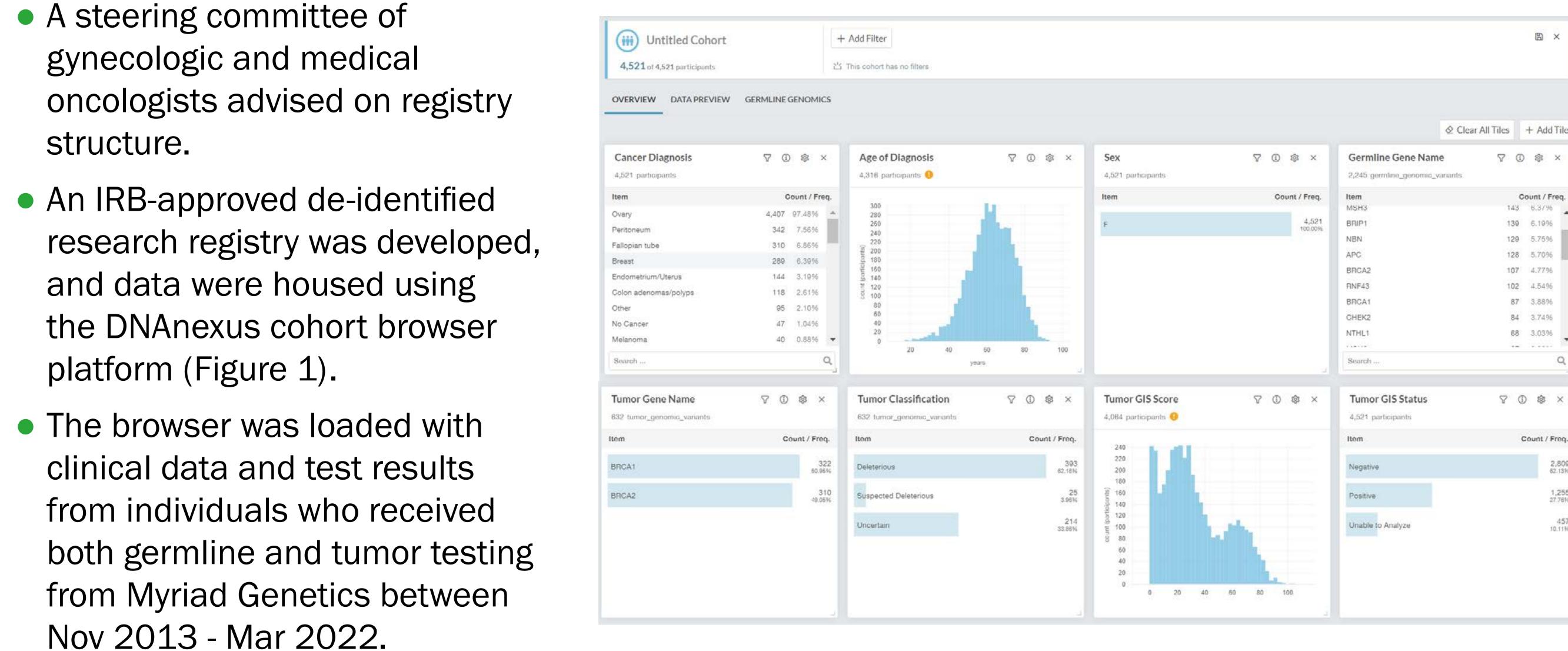
^aDoes not include patients who received germline testing only

^bCategories are not mutually exclusive. Number of cases reported include primary, secondary, and other cancers.

^cSome patients had more than one germline variant.

METHODS

Development Figure 1. Registry Browser Interface A steering committee of



Statistics

structure.

platform (Figure 1).

- Descriptive statistics were used to summarize registry characteristics and access-requesting user demographics as of 1/23/23.
- Reasons for requesting access to the database were evaluated using thematic analysis.

CONCLUSIONS

- In partnership with a steering committee and DNAnexus, we successfully launched a cloudbased registry to advance realworld insights surrounding cancer genomics and treatment.
- Access interest spanned a broad range of clinicians with varying research interests.
- While the majority of the registry currently comprises ovarian cancer cases, future versions will include data from patients with all solid tumors and will incorporate thematic desires of users.
- The registry will also be populated with patient management and outcomes over time.
- This registry has the potential to be an invaluable tool for facilitating high-quality, multifaceted research in the field of real-world precision medicine.

to learn more and to sign up for the registry

