In search of lost variants: How EHR integration can improve genetic medicine

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VANDERBILT UNIVERSITY MEDICAL CENTER

A diagnostic dilemma

- ✓ De novo
- ✓ Frameshift mutation
- ✓ Rare
- ! Not a known disease-causing gene
- ? EHR search for “gene matching individuals”?

UDN patient: 26 year old female

Genetics	De novo *MSL2* c.694_697del, p.Ser232Thrfs*10
Behavior	- Autism - Obsessive compulsive behavior
Learning	- Mild ID (IQ of 69)
Hypermobility	- Hyperextensible joints - Patellar subluxation
Vision	- High myopia

EHR based genetic matchmaking

Undiagnosed Diseases Network

String search for "MSL2"

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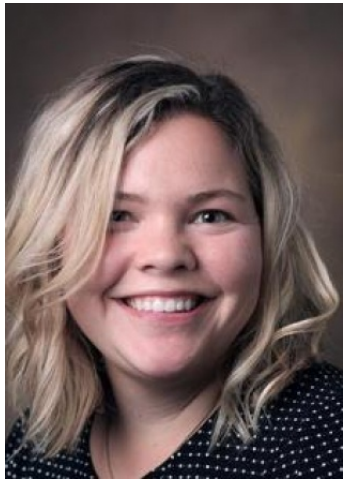


Elly Brokamp

What we want

Clinical genetics database

- Genes tested
- What was found?
  - Gene
  - Variant (HGVS)
  - Zygosity
  - Interpretation
  - Etc.

What we have

PDF genetic reports…

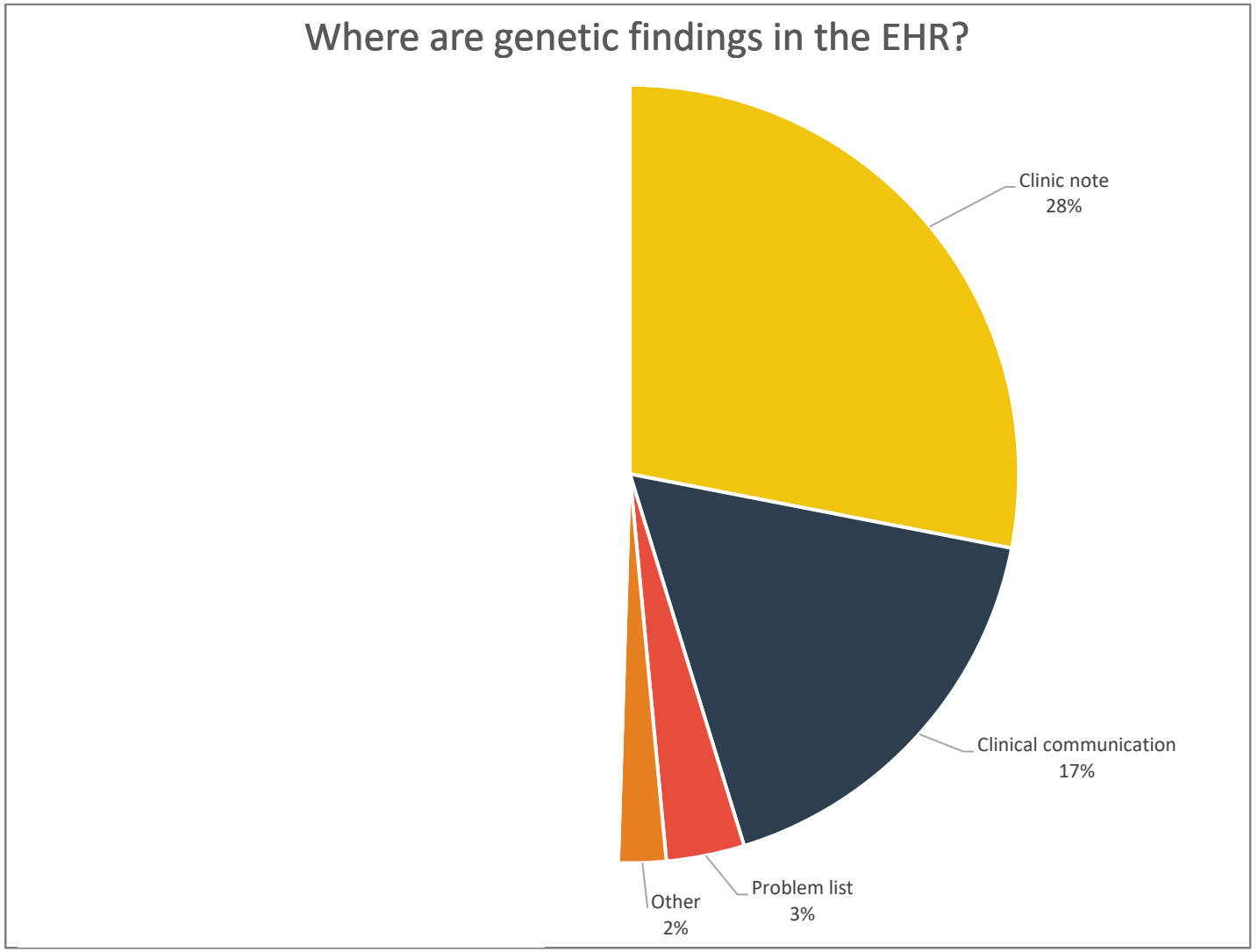
Not part of research databases!

Entered in chart as free text…. sometimes

Gene testing through **Invitae** revealed a **mutation** in **R891Q**-**MYBPC3**, variant of unknown significance.

Incomplete capture of genetic testing results

Indexing medical records by genetic findings



Why I ❤️ clinical genetic variants… and you should too

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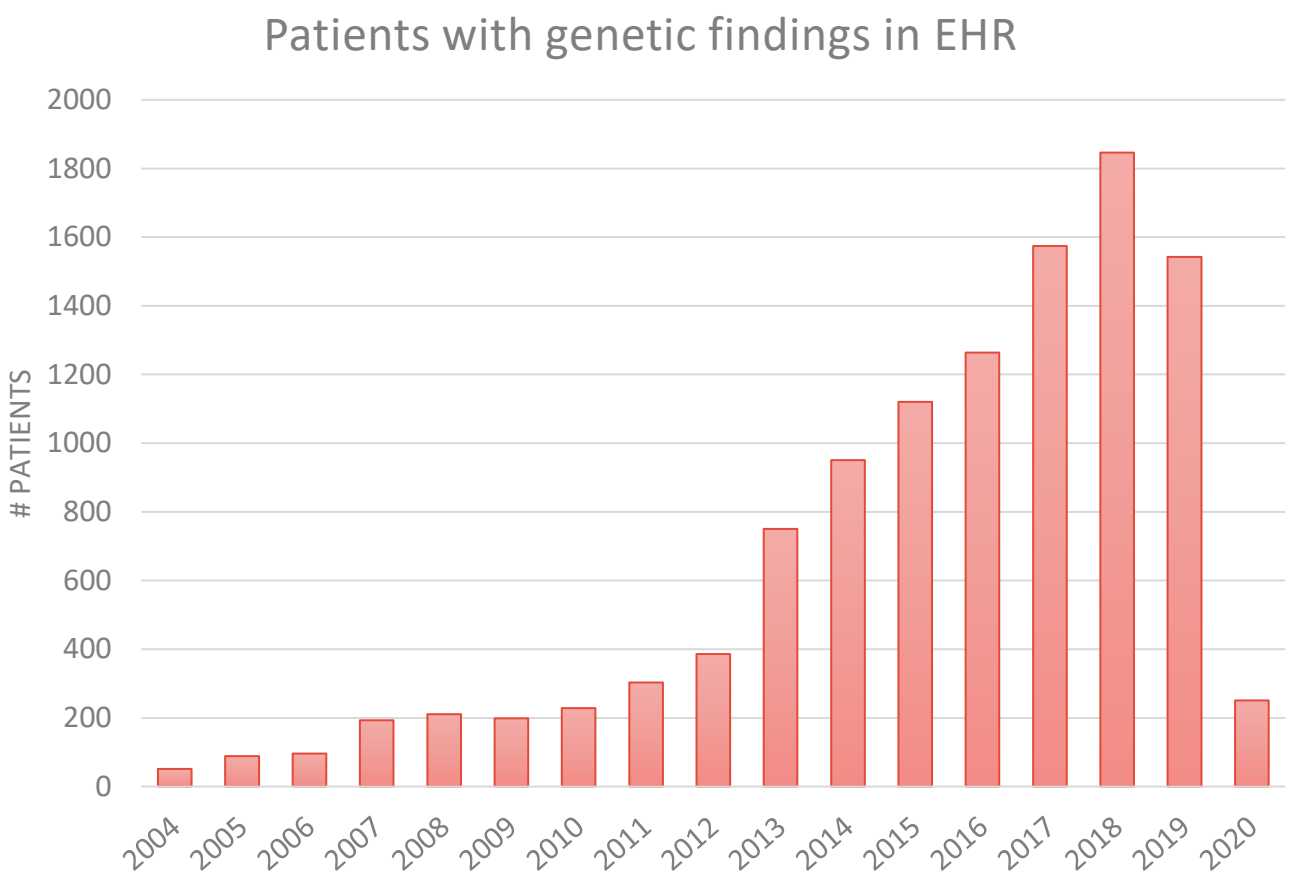
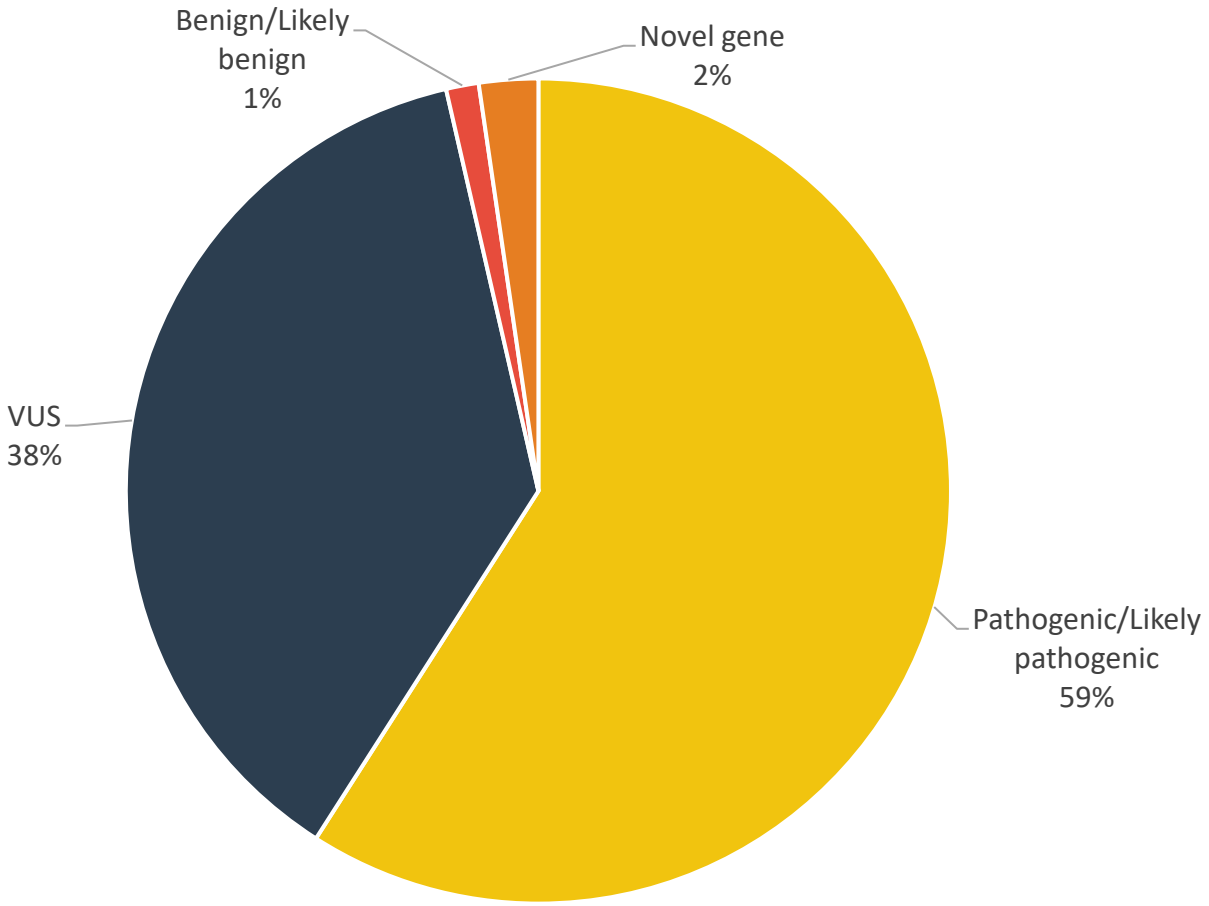
- There are more each day



Why I ❤ clinical genetic variants… and you should too

- They are free
- There are more each day
- They are consequential



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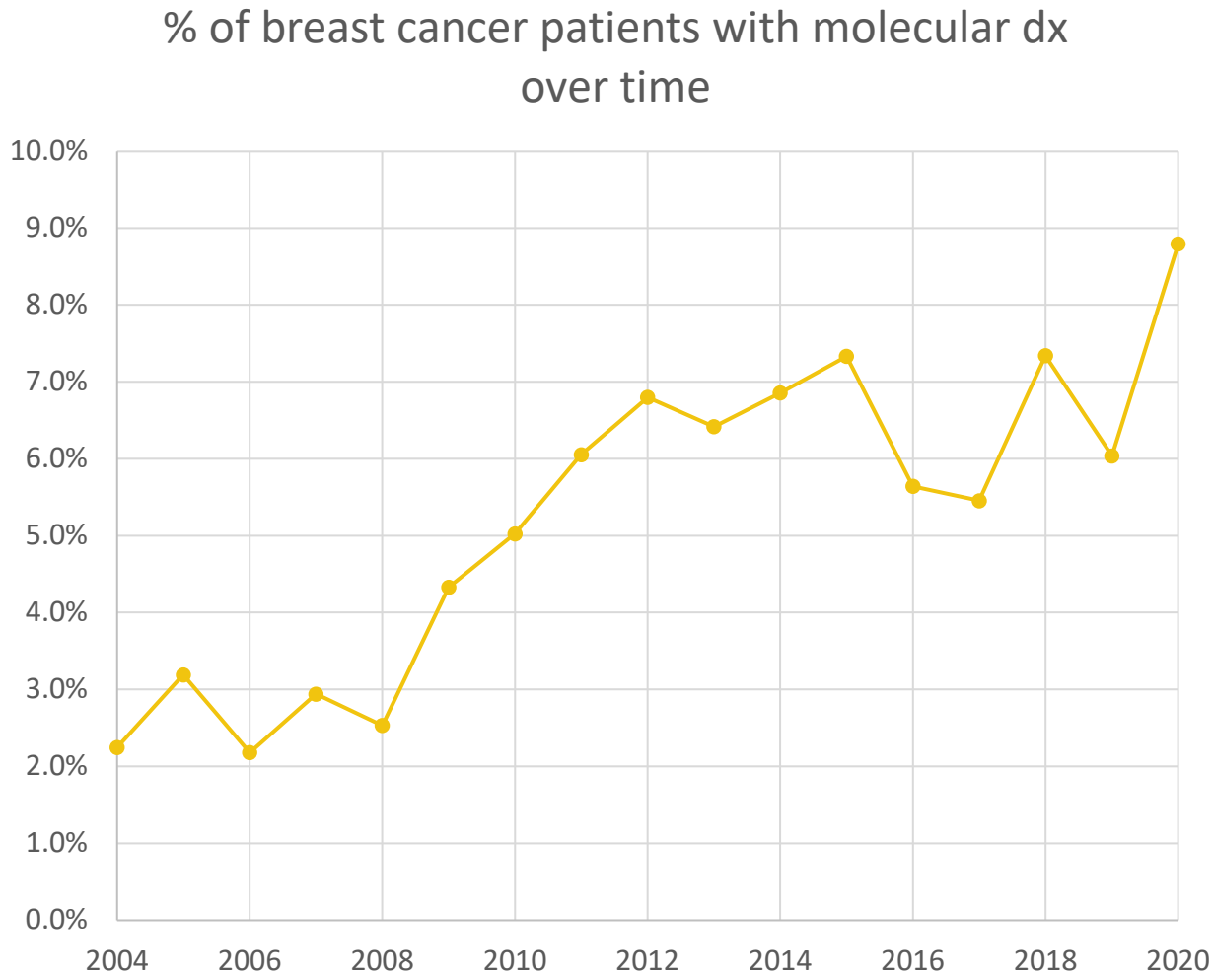

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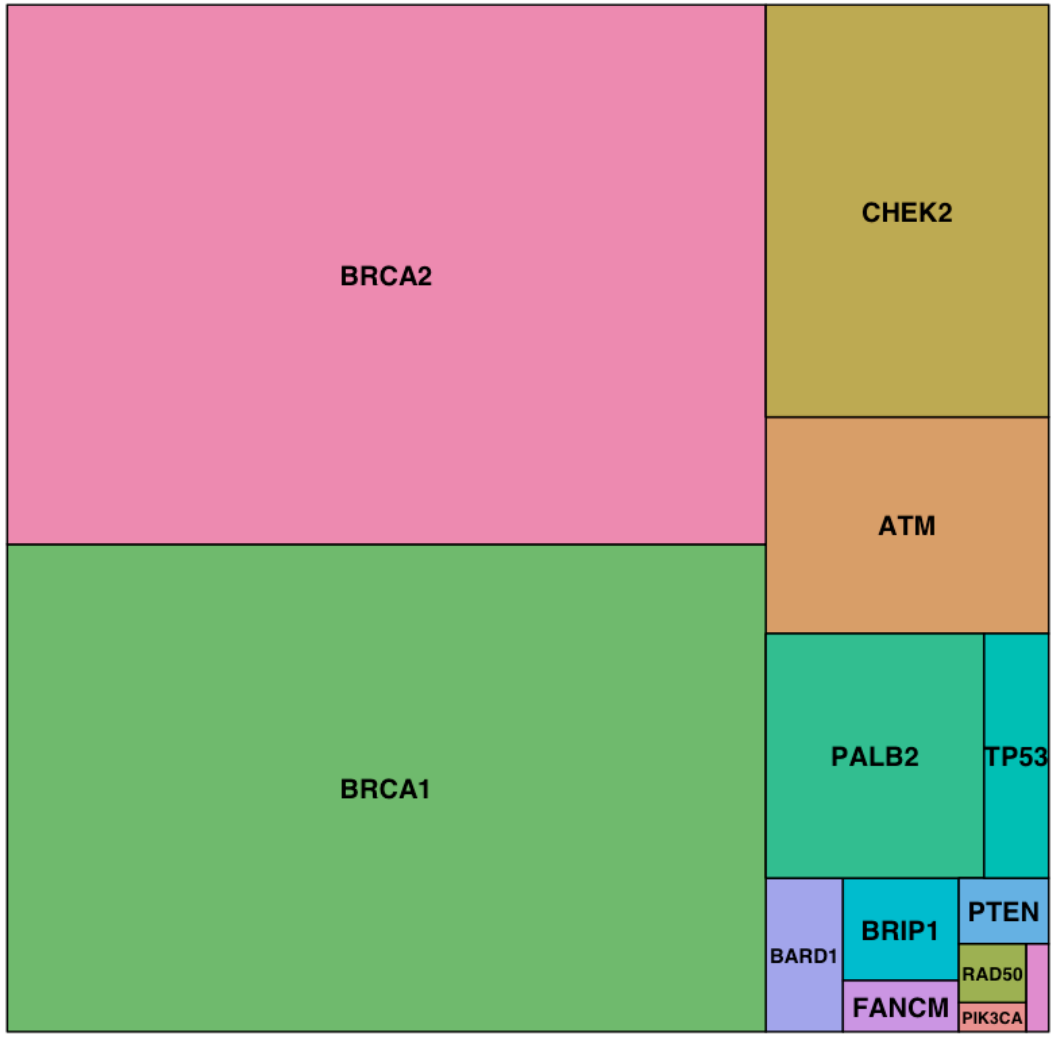
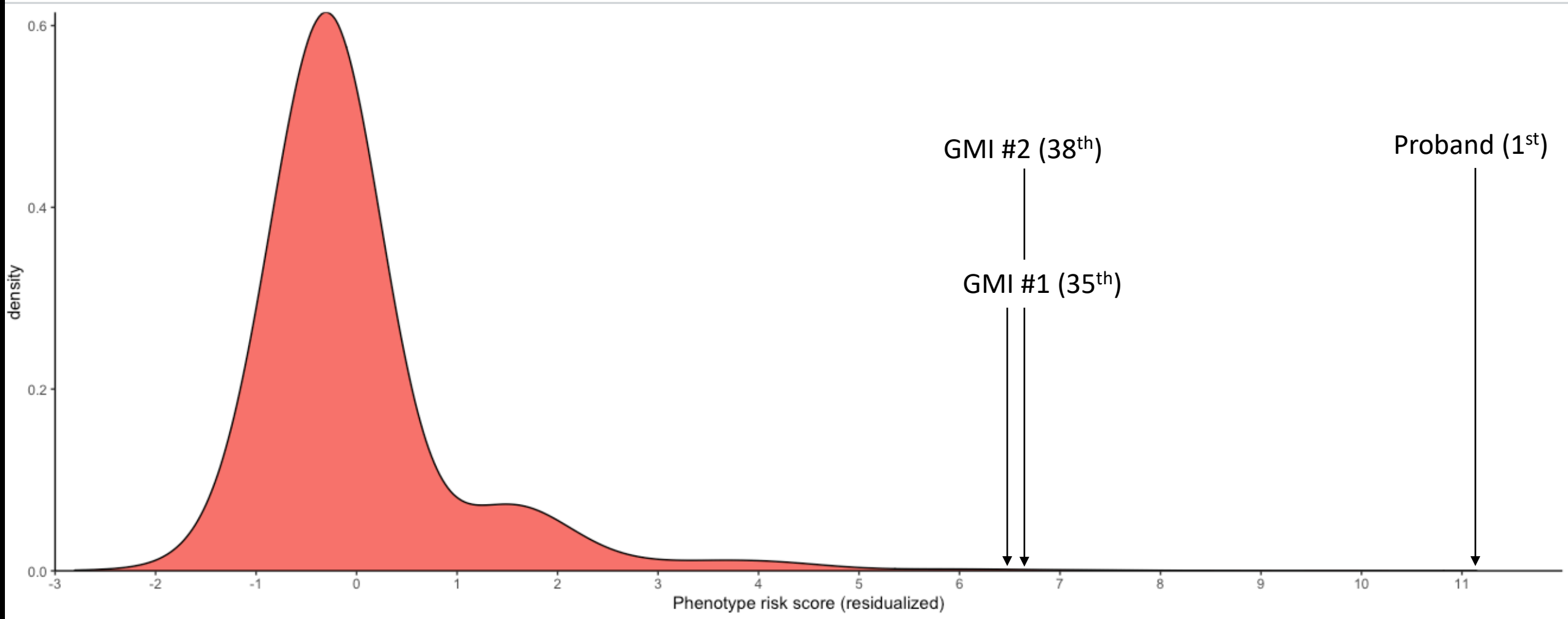
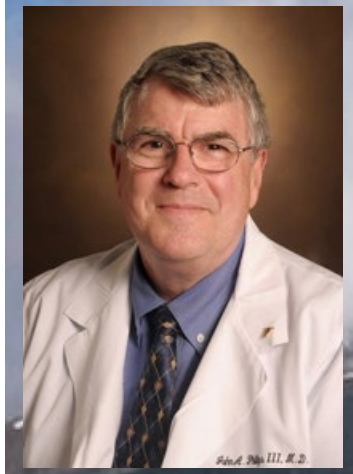
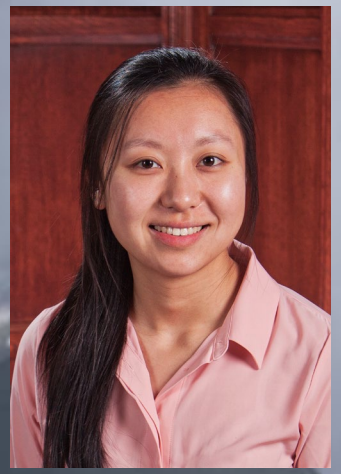
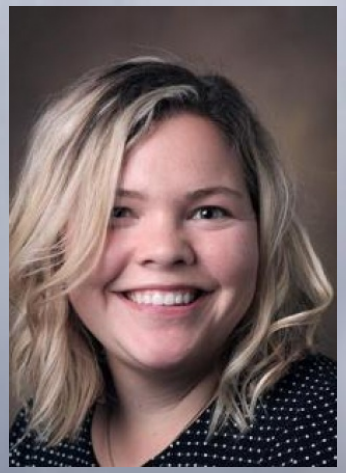


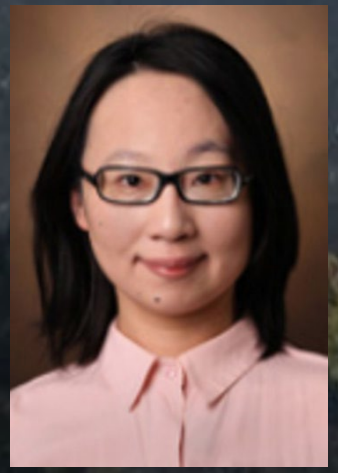
Genes underlying breast cancer molecular dx

Phenotype risk score for *MSL1*







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emerge network
ELECTRONIC MEDICAL RECORDS AND GENOMICS

The time to develop methods is now!

Undiagnosed Diseases Network