Working with DTCGT in the context of research

Andy Singleton
Thursday, October 31, 2019

2019: Meta 5

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New Results

Parkinson's disease genetics: identifying novel risk loci, providing causal insights and improving estimates of heritable risk.

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Mike Nalls

NIH National Institute on Aging *Intramural Research Program*

iPDGC International Parkinson Disease Genomics Consortium

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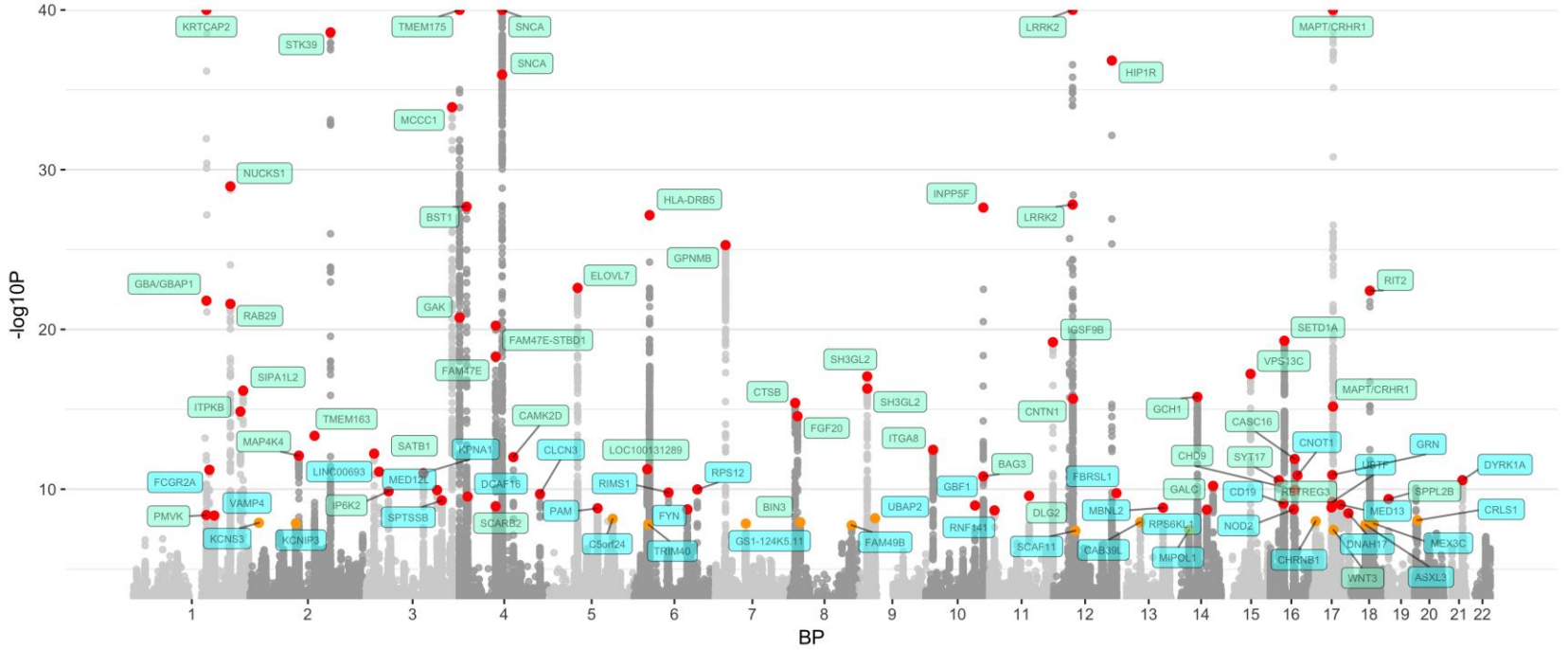
Largest meta-analysis of GWAS data to date:

- meta-analysis and conditional analyses
- 37,688 cases (IPDGC+23andMe)
- 18,618 proxy-cases (UKBB)
- 1,417,791 controls (IPDGC+UKBB+23andMe)
- 11,477,547 imputed SNPs (HRC)



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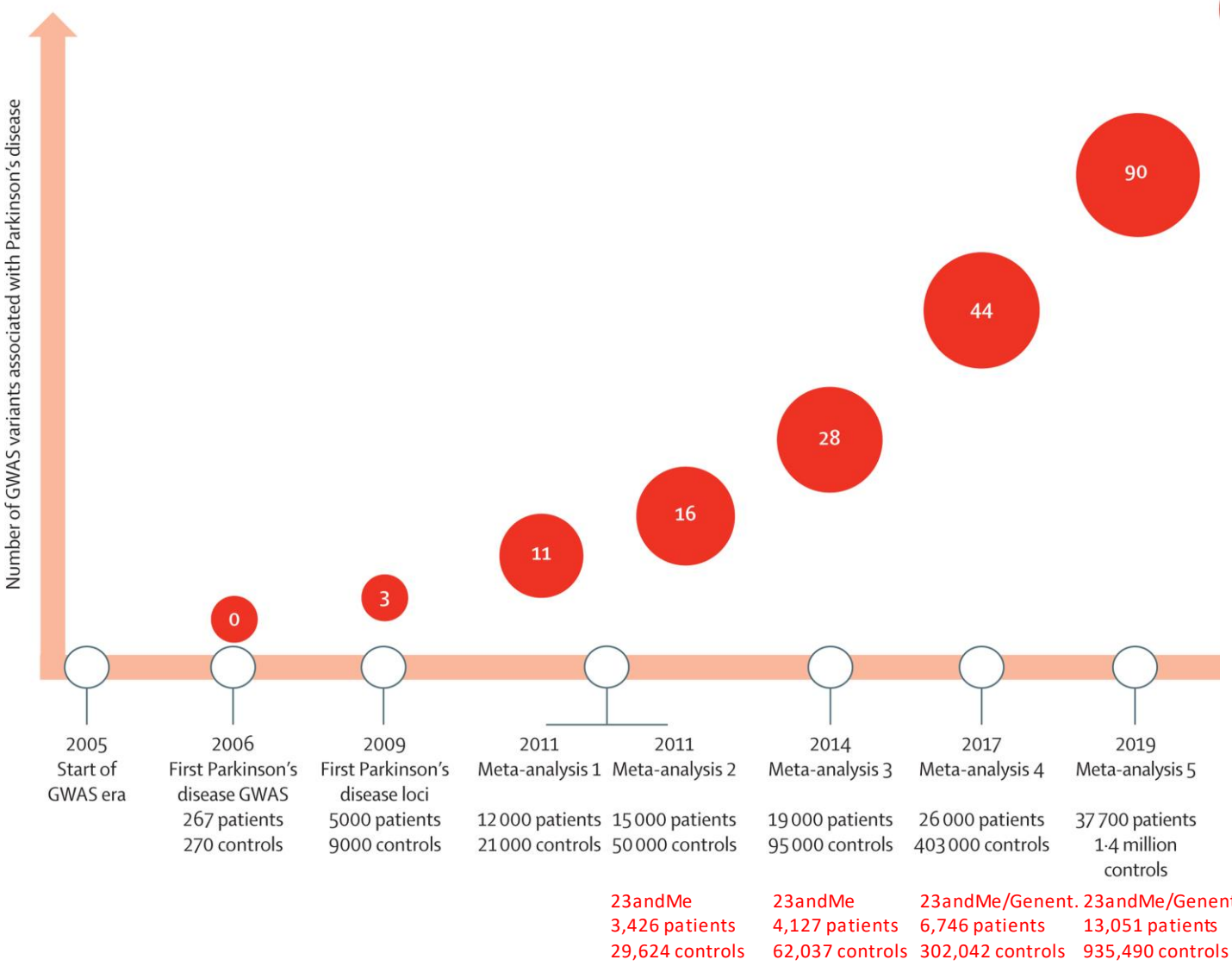

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Our experience

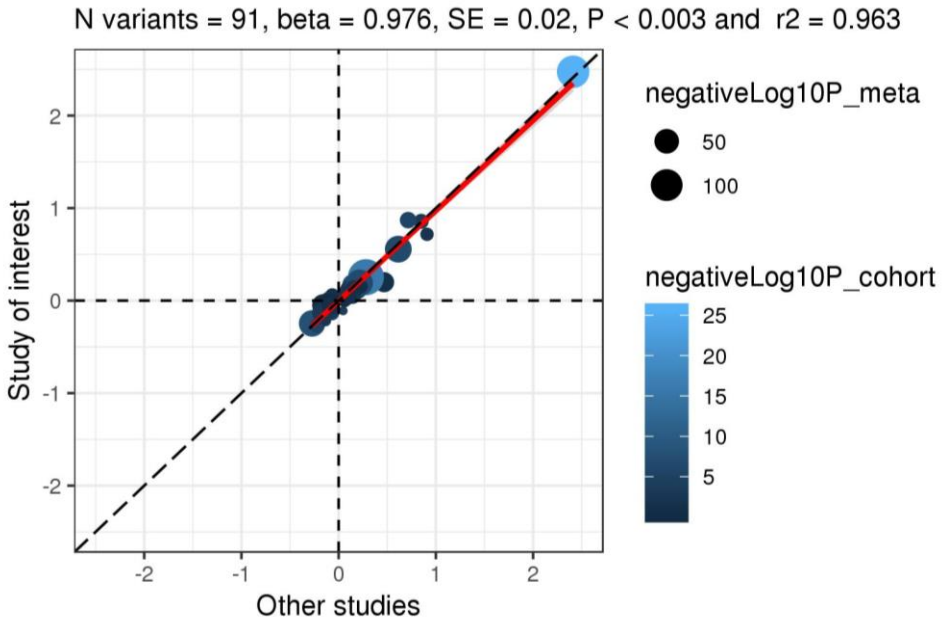
- somewhat unusual because of the subject and the companies interest
- expands beyond simple searches for risk loci for disease
  - related conditions (Mendelian randomization)
  - behaviors that influence disease risk
  - disease variability
  - sex based variation
  - bespoke replication

Strengths and Challenges

- How good are the data

Strengths and Challenges

- How good are the data
  - genetically – very good
  - phenotypically – not really very different than other clinical studies



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Strengths and Challenges

- Speed
  - In general – as responsive as other research groups.
  - There is a process, but the process works well.
  - Note, our area is perhaps an exception. Foundation/philanthropic support has been critical.
- Decisions
  - The company has its own research priorities.
  - Their own target pipeline and/or agreements.
  - Work has to be non-competitive in that regard

Strengths and Challenges

- Analysis and QC – some barriers to what we can do
  - can look for overlap (checksums)
  - some data types not easily accessible (dosage)
- Sharing
  - the biggest challenge – the current policy limits broad data sharing
  - we compartmentalize sharing of results – own data sum. stat., available to download, DTCGT results, require separate agreement
- Scale
  - very very good. The capacity to include proxy cases is also substantial

One Future?

FOX INSIGHT

YOUR EXPERIENCE FUELING RESEARCH

Share Your Expertise

No one understands Parkinson's better than those living with it every day. By working together, we can help shape the future of research.

Participate Online

Fox Insight easily collects self-reported data about health experiences from those with and without Parkinson's in a number of ways.

Drive Genetic Insights

Eligible individuals can help researchers gain a holistic picture of the disease by participating in a genetic sub-study powered by 23andMe.

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Share Data

Join LunaDNA and share your data in a few steps. Your data is de-identified and encrypted to ensure privacy and security.

Help Discovery

Researchers pay to conduct research on the aggregated data.

Earn Benefits

Proceeds earned from research on the platform are passed back to the shareholders who shared their data.