Simons Variation in Individuals Project

DYRK1A Family Meeting Update

July 14, 2018

Simons VIP Overview

- What is Simons VIP?
- What are the steps?
- What do we collect?

Simons Variation in Individuals Project

Online Registry

Recontactable cohort

Community

SimonsVIP**connect**.org

Gene-specific resources

Webinars

Facebook Groups by Gene

Data Repository

Genetic Diagnosis

Lab reports

Phenotypic information

Medical History

Online & phone measures

Longitudinal follow-up

Imaging data

Biospecimen Repository

Whole blood DNA

Saliva DNA

Lymphoblastoid cell lines

Fibroblasts

iPSCs

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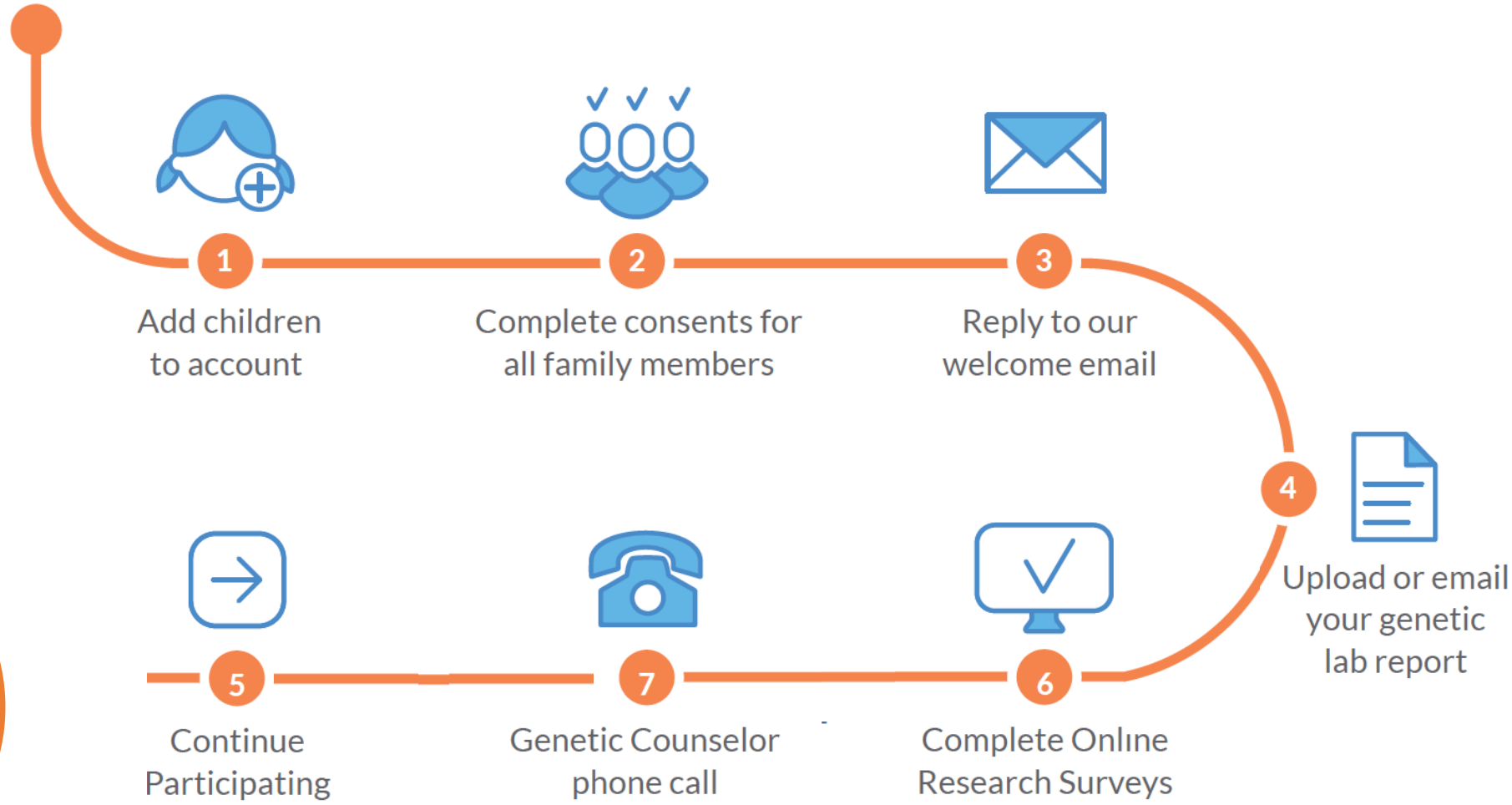
Study Participation Path



Total Consented Participants: 3,409

Total Consented Carriers: 1,180

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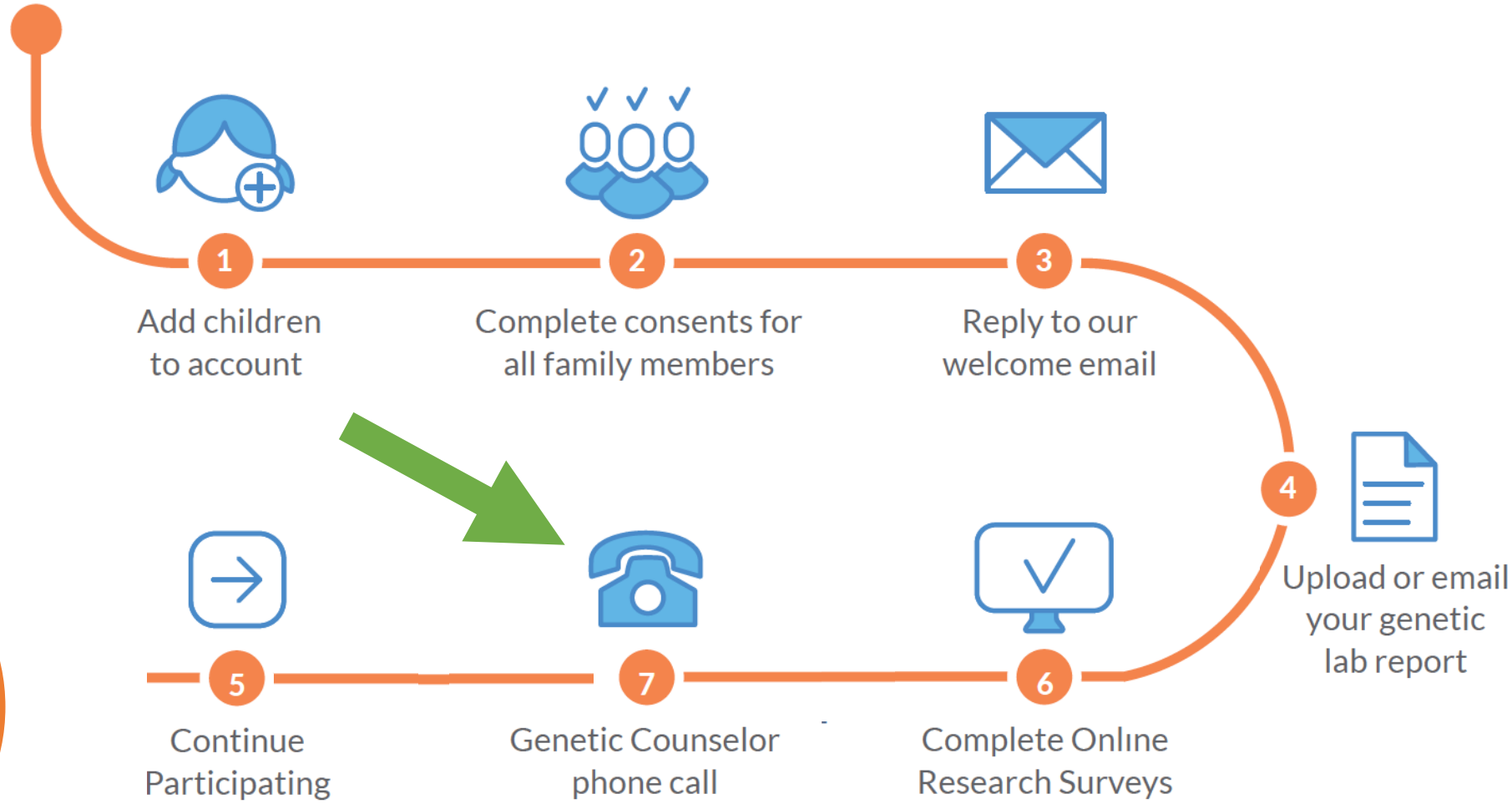
Study Participation Path



Total Consented Participants: 3,409

Total Consented Carriers: 1,180

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What survey data do we collect?

- Clinical Genetic Lab Results
- Medical History Interview (phone)
- Adaptive Functioning Interview - Vineland (phone)
- Seizure Survey
- Background History
- Child/Adult Behavior Checklist
- Social Responsiveness Scale
- Social Communication Questionnaire

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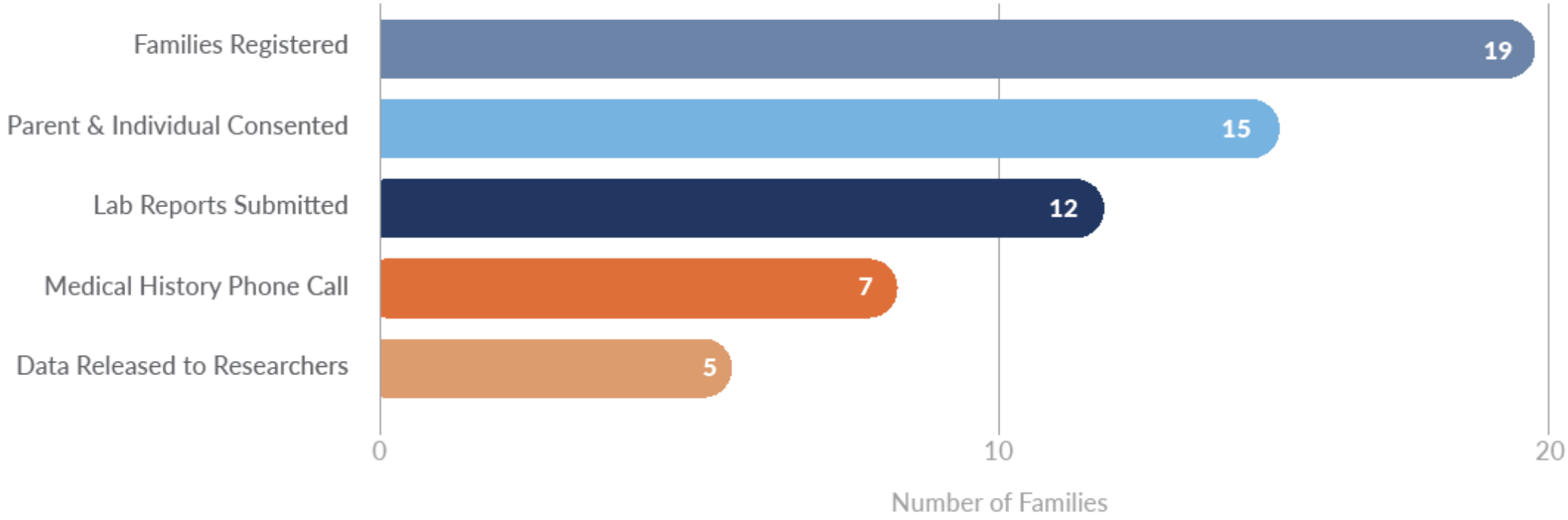
Where are *DYRK1A* families in this process?

Finish any missing steps to contribute your family's data!



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Simons VIP DYRK1A Participant Data

Age range
3yrs - 24yrs

14 / 16
Participants under 18

Average age
11 yrs

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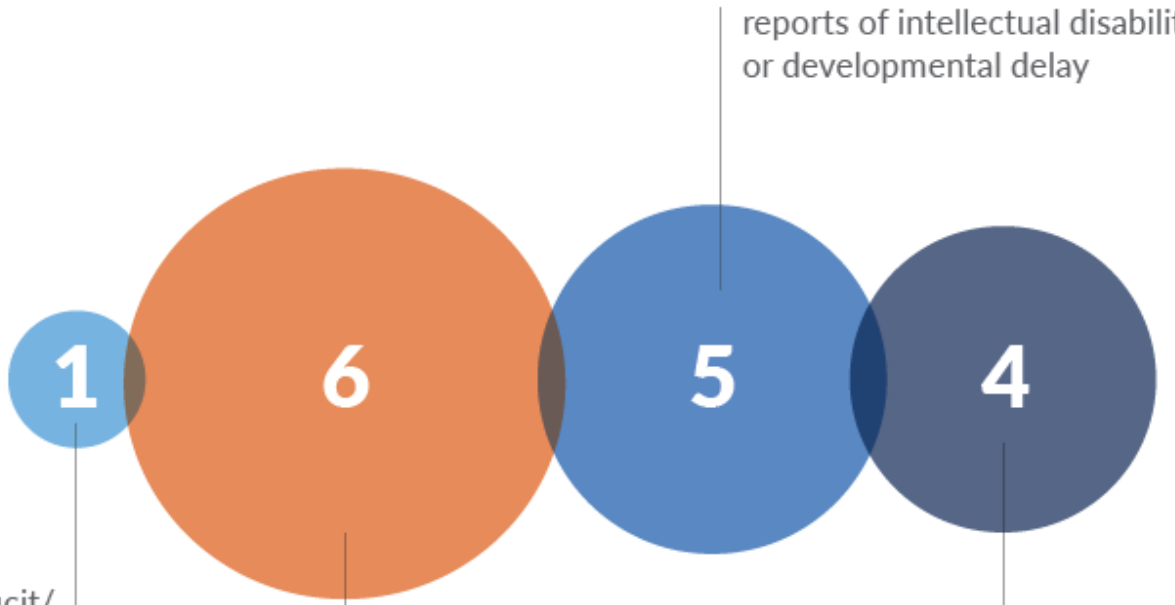
Developmental & Behavioral Diagnoses

Simons VIP completed diagnostic history interviews with 7 participants.



reports of attention-deficit/ hyperactivity disorder

reports of language disorder or delay

reports of intellectual disability or developmental delay

have been diagnosed with autism spectrum disorder

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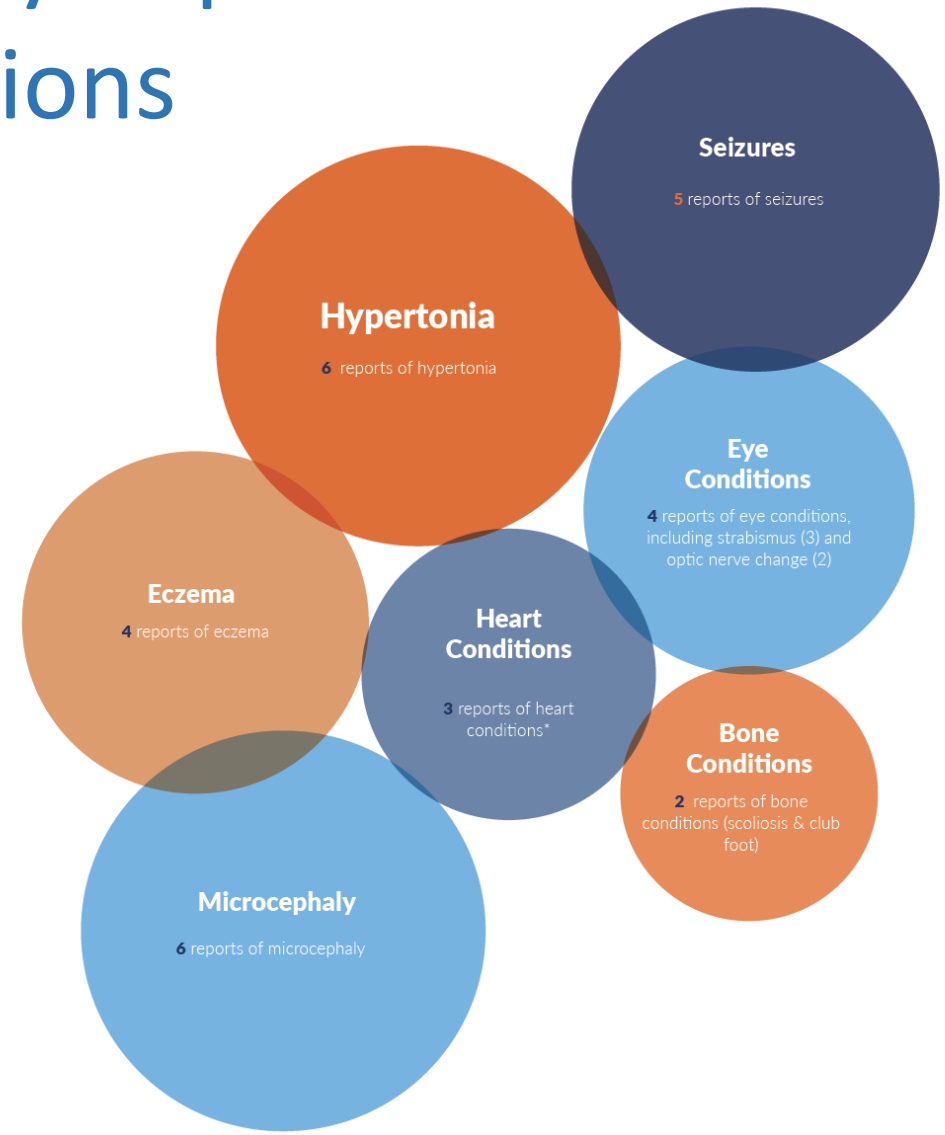
Most Commonly Reported Medical Conditions



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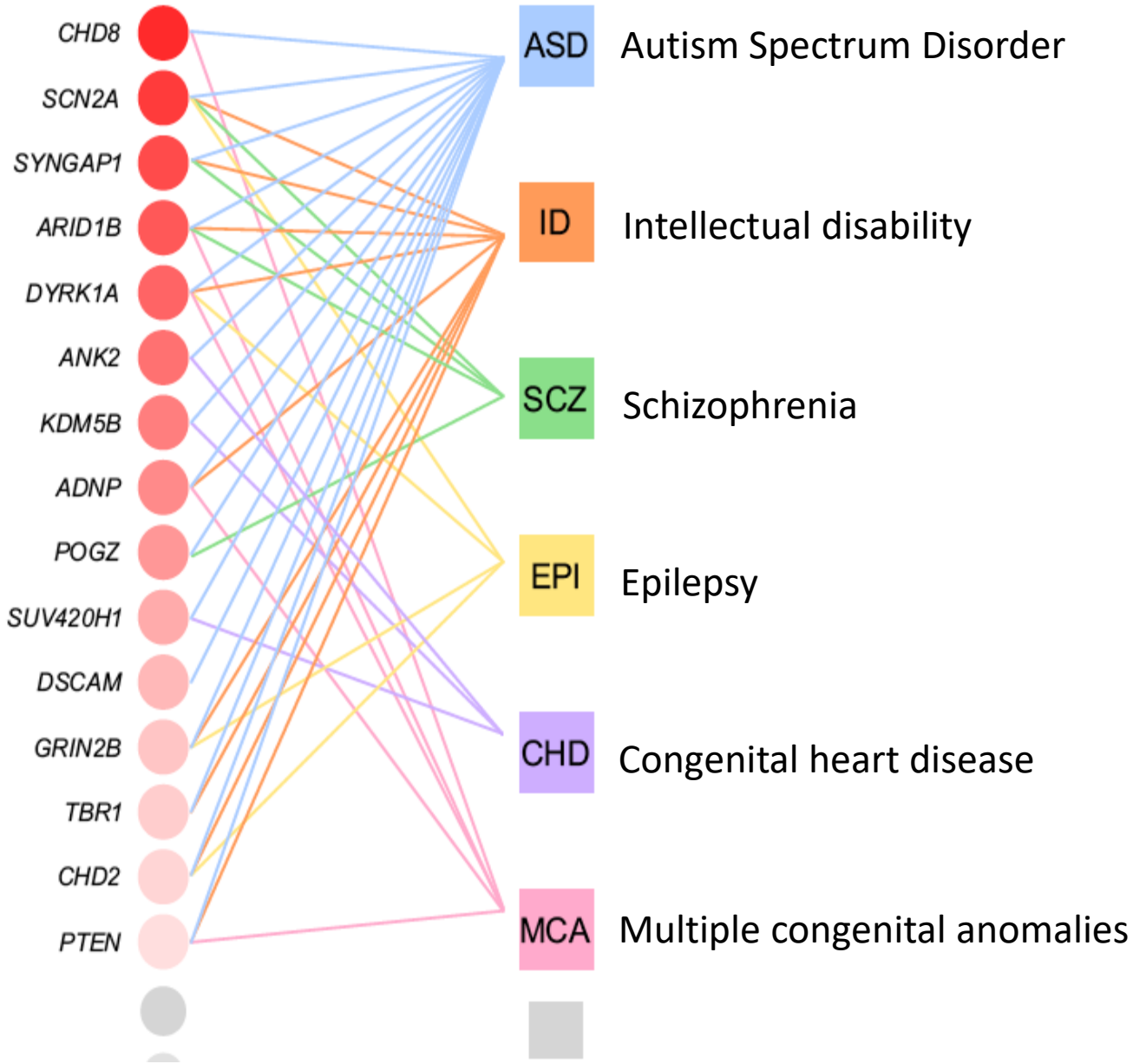
Why participate in a registry?

- Why “Genetics First”?
- External stakeholder needs
- Numbers matter

Genetics First



Buxbaum, Biological Psychiatry May 1, 2015; 77:766–768

External Stakeholder Needs

Researchers

- Accessible
- Common data elements
- Longitudinal
- Ways to merge existing datasets
- Flexible & expandable

Pharma

- Single access point for multiple registries
- Standardized data
- Minimal missing data
- Comprehensive and condition specific
- Include adverse event & medication data

Genes We Study

Copy Number Variants	
16p11.2 deletions	16p11.2 duplications
1q21.1 deletions	1q21.1 duplications

Genes Associated with Features of Autism				
ACTL6B	CHD8	GRIN2A	KMT5B	SCN2A
ADNP	CSNK2A1	GRIN2B	MBD5	SETBP1
AHDC1	CTBP1	HIVEP2	MED13L	SETD5
ANK2	CTNNB1	HNRNPH2	PACS1	SMARCA4
ANKRD11	CUL3	KAT6A	PBRM1	SMARCC1
ARID1B	DDX3X	KATNAL2	POGZ	SMARCC2
ASH1L	DNMT3A	KCNQ2	PPP2R5D	STXBP1
ASXL3	DSCAM	KDM5B	PTCHD1	SYNGAP1
BCL11A	DST	KDM6B	PTEN	TBR1
CHAMP1	DYRK1A	KMT2C	PURA	
CHD2	FOXP1	KMT2E	REST	

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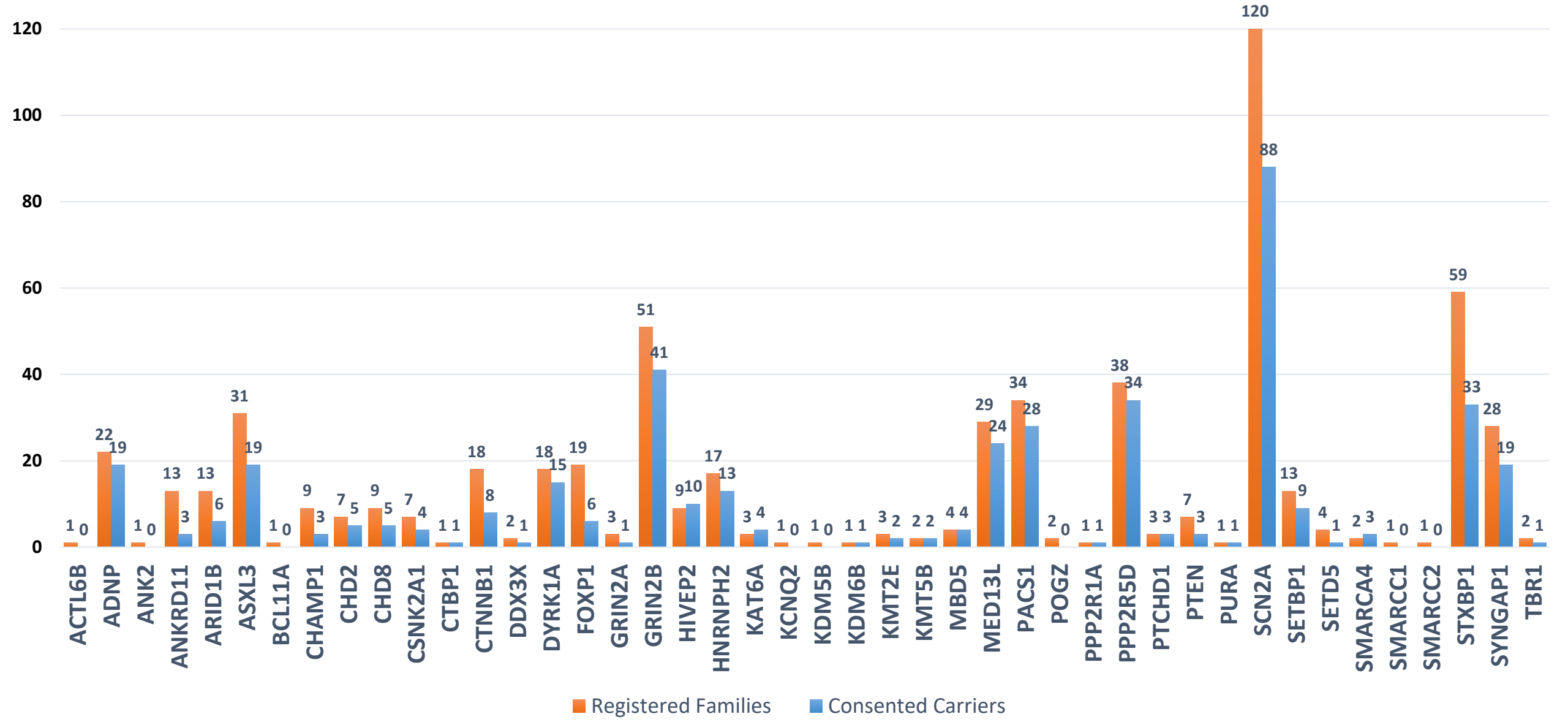
Single Gene Variants: Families Registered & Carriers Consented



Comparing Research Projects

- Simons VIP
- TIGER
- SPARK

Key Distinctions

	SIMONS VIP Simons Variation in Individuals Project	SPARK Igniting autism research Improving lives	TIGER
ASD diagnosis required		✓	
Confirmed genetic diagnosis required	✓		✓
Online data registry	✓	✓	
Phone assessments	✓		✓
Detailed In-person assessments			✓
EEG			✓
Data and samples ordered by outside researchers	✓	✓	*
Participant recontacting for additional projects	✓	✓	✓
Available in other languages	✓		

The Simons VIP Team

Simons Foundation


Chung


Spiro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Cartner

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Taylor


Lese Martin


Faucett


Kasparson


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Singer


Martin


Dent


E. Smith


Weiss


Lehman

Columbia


Wilson

Tempus

Invitae

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Questions?