

## Table 1. Biospecimens and Genetic data:

\*Not all participants have all sample types available. Samples on our website are updated regularly.

Samples*	Web Link
Induced Pluripotent Stem Cells	<a href="https://www.sfari.org/resource/ips-cells/">https://www.sfari.org/resource/ips-cells/</a>
Fibroblasts, Lymphoblastoid Cell Lines, Saliva DNA, Cell Line DNA, Whole Blood DNA	<a href="https://www.sfari.org/resource/simons-searchlight/">https://www.sfari.org/resource/simons-searchlight/</a>

## Table 2. Data types available (phone interview and online surveys):

**Simons Searchlight (formerly SimonsVIP)** involves the collection of family medical, developmental and behavioral information through online surveys and phone interviews with families. All families receive surveys that are part of the standard battery. Additional surveys that are more specific to comorbidities with certain genes (such as the seizure survey) are assigned to participants based on relevance. Some children have completed repeated longitudinal measures semiannually (prior to 2016 for children under age 7) or annually (child carriers over age 7), including Medical History, Vineland and diagnostic updates. More details on these measures can be found on [base.sfari.org](http://base.sfari.org).

**KEY:** \* = Only if adult sibling  
 ~ = If autism spectrum disorder suspected  
 # = Only participants that go on to the functional imaging stage  
 Gray (O) = No longer collected past 2016, earlier for some measures

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier
<b>Medical history</b>				
Medical History Interview (MHI — Adult and Child version)	X		X	
Medication Interview	X		X	
Previous Diagnosis Interview	X	O	X	X
Puberty Questionnaire (versions for boys, girls, men, women)	X		X	
Seizure Survey- Online survey version	X		X	
Simons Searchlight Seizure History			X	
Skin and dental questionnaire			X	
<b>Development</b>				
Vineland Adaptive Behavior Scales-II (VABS-II; interview)	X	O*	X	O
Learn to read questionnaire			X	
<b>Behavior</b>				
Adult Behavior Checklist (ABCL)	X	X*		
ARI (Adult Routines Inventory)	X	X		

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier
<i>Behavior and Stereotyped Interests Questionnaire (BSIQ; Online Version)</i>			○	○
<i>Broader Autism Phenotype Questionnaire (BAPQ)</i>	○	○		
Child Behavior Checklist for ages 6 to 18 years (CBCL)			X	X
<i>Childhood Routines Inventory-Revised (CRI-R)</i>			○	○
<i>GO-LIFE (Adult)</i>	○	○		
<i>GO-LIFE — Child</i>	○			
Modified Checklist for Autism in Toddlers (MCHAT)			X	X
Social Communication Questionnaire—Lifetime (SCQ)				X
Social Responsiveness Scale (SRS-2) (School age 4-18; Adult 19+)	X	X	X	X
<i>Teacher Rating Form (TRF- 1.5-5, 6-18)</i>			○	○
<i>Teacher SRS-2</i>			○	○
<b>Demographics</b>				
Simons VIP Background History Form	X		X	

**Table 3. Phase 1 in-clinic study for 16p11.2 and 1q21.1 deletions and duplications (Phase ended early in 2014)**

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
<b>Neuropsychological Assessments</b>						
Autism Diagnostic Interview-Revised (ADI-R)	X~	X*~	X~	X~		
Autism Diagnostic Observational Schedule (ADOS)	X	X*	X	X		
Vineland Adaptive Behavior Scale-II (VABS-II)	X	X*	X	X		X
Autism Diagnostic Observational Schedule for Toddlers (ADOS-T)			X	X		
Cognitive Testing	X	X	X	X	X	X
Delis-Kaplan Executive Function System (DKEFS)	X		X		X	X
Purdue Pegboard	X	X	X	X	X	X
Movement Assessment Battery for Children-Second Edition (Movement ABC-2)			X			
Clinical Evaluation of Language Fundamentals (CELF-4)	X#		X#		X	X

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
CASL (Comprehensive Assessment of Spoken Language)			X	X		
CCC-2 (Children's Communication Checklist-2)			X	X		X
Comprehensive Test of Phonological Processing — Non-Word Repetition task (CTOPP)	X	X	X	X	X	X
MacArthur Communicative Development Inventory — Words and Gestures, and Words and Phrases versions (MCDI)			X	X		
WIAT III (Wechsler Individual Achievement Test, Version III)	X		X		X	X
<b>Behavior</b>						
Adult Behavior Checklist (ABCL)	X	X*			X	
Child Behavior Checklist for ages 6 to 18 years (CBCL)			X	X		X
Diagnostic Interview Schedule for Children (DISC) — Young Child and Child versions			X	X		X
Symptom Checklist-90-Revised (SCL-90-R)	X	X			X	
Broader Autism Phenotype Questionnaire (BAPQ)	X	X			X	
Behavior and Stereotyped Interest Questionnaire (BSIQ)			X	X		X
Parenting Stress Index (PSI)			X	X		
SCQ (Social Communication Questionnaire—Lifetime)			X	X		X
Social Responsiveness Scale (SRS)			X	X		X
Social Responsiveness Scale-Adult Research Version (SRS-ARV)	X	X			X	
<b>Medical and other history</b>						
Dysmorphology—3dMD Pictures	X	X	X	X		
Education History Interview	X		X			
Family Structure Form (Pedigree)	X		X			
Intervention History Interview	X		X			
Medical History Interview (MHI — Adult and Child versions)	X		X			
Medication Interview	X		X			
Mother Other Pregnancy Questionnaire	X	X				
Previous Diagnosis Interview (Parent report on child, Self-Report Versions)	X	X				
Puberty Questionnaire (versions for boys, girls, men, women)	X	X	X			

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
Simons VIP Background History Form	X		X		X	X
Simons VIP Feeding Questionnaire			X	X		X
Eating in Absence of Hunger — Adult Self-Report, Parent Report on Child, and Child Self-Report versions (EAH)	X	X	X	X	X	X
LOC (Loss of Control) — Parent report on child, Self-report versions	X	X	X	X	X	X
Pediatric Sleep Questionnaire			X	X		X
Adult Sleep Questionnaire	X	X			X	
Edinburgh Handedness Inventory	X	X	X	X	X	X
HWHC Form (Height, Weight, Head Circumference Form)	X	X	X	X	X	X
Neurological Exam	X	X			X	X
Neurologic Record Review Form (NRRF)	X	X				
Structural MRI Scan Results Form	X	X*	X	X		