

SFARI Base
SSC/Simons VIP/SPARK
Researcher Welcome Packet

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Table of Contents

SSC PARTICIPATION CRITERIA	1
Participants in the Simons Simplex Collection	1
Inclusion Criteria	1
Exclusion Criteria	2
Other Considerations	4
SSC@IAN FOLLOW-UP STUDY PARTICIPATION CRITERIA	6
Inclusion Criteria	6
Exclusion Criteria	6
SIMONS VIP PARTICIPATION CRITERIA	7
Phase 1 Inclusion Criteria	7
Phase 1 Exclusion Criteria	7
Phase 2 Inclusion Criteria	10
Phase 2 Exclusion Criteria	11
SPARK PARTICIPATION CRITERIA	12
Inclusion Criteria	12
Exclusion Criteria	12
SSC AVAILABLE INSTRUMENTS BY FAMILY ROLE TABLE	13
SSC@IAN FOLLOW-UP STUDY AVAILABLE INSTRUMENTS BY ROLE TABLE	20
SIMONS VIP AVAILABLE INSTRUMENTS BY ROLE TABLE	23
Simons VIP Phase 1: In-clinic study measures	23
Simons VIP Phase 2: Online study measures	34
SPARK AVAILABLE INSTRUMENTS BY FAMILY ROLE TABLE	38

PREREQUISITES FOR ACCESS TO SFARI BASE	39
Required Institutional Documents	39
ONLINE SFARI APPLICATION INSTRUCTIONS	40
SIMONS SIMPLEX COLLECTION SAMPLE CONSENT FORM	41
SIMONS VIP SAMPLE CONSENT FORM	41
Phase 1	41
Phase 2	41
SPARK SAMPLE CONSENT FORM	41

SSC Participation Criteria

Participants were admitted into the SSC in biological family groups. Each family includes one (and only one) child with autism. All siblings must be unaffected.

Participants in the Simons Simplex Collection

Proband: the affected child in a family

Parents: biological mother and father of the proband, living at the time of submission, both unaffected with autism — The parents must not be related to each other by blood.

Unaffected designated sibling: the unaffected full sibling closest in age to the proband — The sibling was at least 4 years of age at the time of biospecimen collection.

Other sibling(s): other sibling, opposite in gender to the designated sibling (see other considerations below) — This sibling was at least 4 years of age at the time of biospecimen collection. *We did not collect blood or data on siblings under age 4.

Sibling exception: full sibling requirement was waived for approximately 20 percent of families due to the following exceptions:

- no sibling available
- only half siblings
- full or half siblings younger than 4 years old

Blood samples were collected from the proband, mother, father and designated (full) sibling and sent to a central repository (Rutgers University Cell and DNA Repository), where biospecimens were generated.

Inclusion Criteria

Age

The proband was between 4 years and 17 years 11 months of age when the phenotype measures were administered and the data were collected.

All measures were completed within a six-month time period for all individuals.

ADI-R Scores

On the ADI-R, the proband met one of the following four (4) criteria:

- standard cutoffs on the social and communication domains

- standard cutoff on the social domain and within two (2) points of communication domain cutoff
- standard cutoff on the communication domain and within two (2) points of social domain cutoff
- was within one (1) point of the standard cutoffs on both social and communication domains

All ADI-R data were collected by a reliable examiner.

ADOS Scores

On the ADOS, the proband received a valid and reliable administration, and met cutoffs for autism spectrum or autism:

- The newest ADOS algorithms were used for Modules 1–3.
- Original cutoff algorithms were used for Module 4.

Nonverbal Cognitive Ability

Using the Mullen Scales of Early Learning, the Differential Ability Scales-II, the Wechsler Intelligence Scale for Children-IV or the Wechsler Abbreviated Scale of Intelligence:

- probands between 4 years and 6 years 11 months of age (48 to 83 months) must have had a nonverbal mental age of at least 24 months. Some exceptions down to 18 months nonverbal mental age were allowed after careful review of diagnosis including observation by videotape.
- probands 7 years of age or older (84 months or older) must have had a nonverbal mental age of at least 30 months.

Diagnosis

Following the SSC evaluation, the proband received a clinical ‘best estimate diagnosis’ of autistic disorder, Asperger’s disorder or pervasive developmental disorder not otherwise specified (PDD-NOS), according to the *Diagnostic and Statistical Manual of Mental Disorders* (DSM-IV-TR).

- The best estimate diagnosis was based on information from the SSC evaluation, including observation, chart review and ADI-R and ADOS findings.
- The best estimate diagnosis was made by a psychologist or physician with appropriate credentials and experience requisite for making clinical psychiatric or psychological diagnoses. The diagnostician had direct observation of the proband in making the best estimate diagnosis.

Exclusion Criteria

Families meeting one or more of the following were excluded:

For Probands

Pregnancy and Birth

- fewer than 36 weeks gestation *and* less than 2,000 grams (approximately 4 lbs., 6.5 oz.) at birth
- born with significant injury, abnormality or disease having effects upon the brain
- a history of extensive prenatal or birth complications:
 - A history of significant prenatal/birth injury or brain damage was explored if the proband was in the neonatal intensive care unit or in the hospital for more than three days after birth.

Other Disorders or Limitations

- positive for fragile X syndrome or Down syndrome (trisomy 21)
 - Note 1: If probands met criteria for inclusion based on clinical phenotype measures, no other genetic diagnosis (whether based on macroscopic, microscopic or sub-microscopic criteria) is excluded, unless features of the syndrome create secondary exclusionary factors (e.g., blindness, intelligence quotient out of range). The interest in these situations was to identify additional risk factors or modifier genes that may correlate with the autism phenotype.
 - Note 2: 'Clinical arrays' may be administered for purposes of genetic counseling, but results from clinical arrays will not be considered in the research project.
- sensory or motor difficulties that would preclude valid use of the diagnostic instruments
- history of severe nutritional or psychological deprivation
- when English is not equal to proband's primary language in use and fluency

For Full or Half Siblings

- diagnosed with, referred for or suspected of having autism
- suspected of having intellectual disability (mental retardation) or severe learning disability
 - exception: cases documented as Down syndrome (trisomy 21)
- diagnosed with schizophrenia
- diagnosed with a developmental or psychiatric disorder
 - *All* developmental and psychiatric disorders, and treatment with psychotropic medication, were potential exclusions and were reviewed by consultants.
 - These siblings were met by a site clinician for behavioral observations to confirm inclusion.
- with an Individual Education Plan (IEP) for extensive special education services
 - Note 1: IEP must be reviewed. If passed, the sibling should be included as the designated sibling and met by the site clinician.
- extensive special education services to exclude
 - any self-contained special education classroom
 - pull-out time of more than 60 minutes per day

- paraprofessional aide assignment
- speech/language therapy for expressive/receptive language or pragmatic language disorder (not articulation)
- occupational therapy for sensory integration
- social skills therapy
- Note 2: For any full siblings less than 4 years old, stringent screening must be done to eliminate any concerns of potential autism. For this reason, if the sibling was receiving any services (even services not considered 'extensive'), the family was put on a wait list until the sibling reached the age of 4. At that time, they could be reassessed for the SSC.

For Biological Parents

- diagnosed with, referred for or suspected of having autism
- suspected of having intellectual disability (mental retardation) or severe learning disability
- diagnosed with schizophrenia
- parents are third-degree relatives or closer
- neither parent is fluent in English

For Second-degree Relatives

Second-degree relatives include aunts, uncles, nephews, nieces, grandparents and grandchildren.

- diagnosed with or suspected of having autism

For Third-degree Relatives

Third-degree relatives include cousins, great-aunts, great-uncles, great-grandmothers, great-grandfathers, half-aunts and half-uncles.

- diagnosed with or suspected of having autism

Participation in Other Biomaterial Repositories

A family was excluded from participating in the SSC if they had participated or were participating in other autism repositories or repositories of disorders related to autism (e.g., obsessive-compulsive disorder).

Other Considerations

Additional Siblings

If an additional sibling opposite in gender to the designated sibling was available, the site collected blood and all designated sibling measures from that sibling.

Multiplex Families

Simplex families initially enrolled in the study may be discovered to be multiplex after the start of data collection.

If a site later identified/identifies that a sibling, parent or second- or third-degree relative is on the autism spectrum (based on additional information from family follow-up or another source), the family will still be available on SFARI Base and flagged as *multiplex* (in the *families_by_distribution* file).

Previously Excluded Families

Families previously excluded from the core SSC collection for various reasons and released in an ancillary set are now available in the main collection and flagged as *Simons Ancillary Collection* (in the *families_by_distribution* and *individuals_by_distribution* files).

Monozygotic Twins with autism

If a family has monozygotic (identical) twins with autism, the proband twin (MZ) protocol in the database was assigned to twin #2 (the proband's twin). The full assessment protocol for the proband was completed on both twins.

SSC@IAN Follow-Up Study

Participation Criteria

Inclusion Criteria

All SSC participants enrolled in the SSC@IAN registry were invited to participate in an online, longitudinal study. For probands and designated siblings who were adults at the time of the study, we first asked a parent to report guardianship status for their adult child (legally independent or dependent). Probands and designated siblings over the age of 18 were invited to participate if they were legally independent. Parents of probands and designated siblings who were minor children or dependent adults were invited to complete surveys about their child(ren). Data were collected on 440 families from the SSC.

Exclusion Criteria

Families in the original SSC who declined to participate in SSC@IAN were not invited to participate in this study.

Simons VIP Participation Criteria

Phase 1 Inclusion Criteria

All participants identified with the specific genetic mutation likely to be involved in autism or other neurodevelopmental disorders were eligible for inclusion in the study, regardless of age, diagnosis or other medical conditions. The Simons VIP started with a focus on deletion (del) and duplication (dup) of chromosomal region 16p11.2, later adding individuals with 1q21.1 deletion or duplication as a small pilot. Inclusion criteria were any individual of any age with the 16p11.2 deletion or duplication, defined as equal to or smaller than 28.5-31.2 megabases. This included individuals with a deletion or duplication of class one, distal 1q21.1 that did not include the TAR region (approximate genomic coordinates 146-147.8 megabases, hg 19).

All measures were completed within a six-month time period for all individuals.

fMRI and MEG Carriers

Participants with a 16p11.2 or 1q21.1 deletion or duplication who were at least 6 years old and demonstrated an ability to remain still during the structural MRI scan done at one of the clinical sites were eligible to continue on to the fMRI/MEG scan phase.

Controls

fMRI and MEG scanning sites recruited matched control participants as part of local site recruitment.

Control-Matching Criteria Guidelines

- age
- gender
- ethnicity
- handedness
- intelligence quotient (IQ) within one standard deviation of the match carrier's IQ
- control's nonverbal IQ within one standard deviation of his/her own verbal IQ

Phase 1 Exclusion Criteria

Carriers

- additional known genetically confirmed mutations with effects on neurocognitive outcome

- 16p11.2 deletions or duplications that are larger than the indicated 2.7 megabase interval; individuals with a deletion or duplication of class one, distal 1q21.1 that include the TAR region or deletion/duplication of just the TAR region.
- carriers and parents who do not speak English fluently — Participants are required to speak English because the behavioral and neuropsychological tests have only been validated and made available in English.
- other early environmental insults that may affect neurocognitive outcome, such as fetal alcohol syndrome, severe birth asphyxia or severe prematurity (gestational age of less than 30 weeks).

Non-carrier siblings

- known genetically confirmed mutations other than 16p11.2 or 1q21.1 with effects on neurocognitive outcome
- siblings who do not speak English fluently — Participants are required to speak English because the behavioral and neuropsychological tests have only been validated and made available in English.
- other early environmental insults that may affect neurocognitive outcome, such as fetal alcohol syndrome, severe birth asphyxia or severe prematurity (gestational age of less than 30 weeks).

fMRI/MEG carriers and controls

- dysmorphic features*
- genetic findings (blood draw and subsequent testing)*
- inability to tolerate MRI scans
- metal implants (e.g., pacemakers)
- dental braces, built-in retainers or space maintainers
 - includes excessive crowns and amalgam (metal) fillings
- pregnancy
- claustrophobia
- obesity (see maximum capacity for MEG and MRI)
- known allergies to fiducial (permanent) markers, medical tape, alcohol wipes
- tattoos on head or neck
- non-removable body piercings
- immediate family members diagnosed with autism or other developmental disorders*
- information about the individual indicating that he or she may have psychiatric or other problems (including significant substance abuse) that may confound interpretation of comparisons — For example, multiple or extremely high elevations on the Broad Autism Phenotype Questionnaire, Symptom Checklist-90, Adult Behavior Checklist/Child

Behavior Checklist, Social Responsiveness Scale or Social Communication Questionnaire, concerning findings from the Dominance, Influence, Steadiness and Conscientiousness assessment, or clinician behavioral observations were considered and reviewed prior to inclusion.*

- 16p11.2 deletions or duplications that are larger than the indicated 2.7 megabase interval; individuals with a deletion or duplication of the class one, distal 1q21.1 region that includes the TAR region.
- controls, carriers and parents who do not speak English fluently — Participants are required to speak English because the behavioral and neuropsychological tests have only been validated and made available in English.
- other early environmental insults that may affect neurocognitive outcome, such as fetal alcohol syndrome, severe birth asphyxia or severe prematurity (gestational age of less than 30 weeks).

* Starred items are for controls only.

Maximum Capacity/Measurements for MEG and MRI:

	Weight	Width (Shoulder)	Head Circumference
MEG	300 lbs.	n/a	62 cm
MRI	400 lbs.	60 cm	64 cm (with padding) 70 cm (without padding)

Phase 2 Inclusion Criteria

All participants of Simons VIP Phase 1 were invited to participate in Phase 2, which was an online extension of the Phase 1 study. Phase 2 participants received a slightly different battery of measures.

Phase 2 expanded on the study, moving to an online-only platform which removed the travel requirement and enabled collection of longitudinal data. Phase 2 permits the rapid integration and study of new groups with recurrent genetic changes other than 16p11.2 and 1q21.1 deletions and duplications. To date, 53 single genes associated with neurodevelopmental differences and features of autism have been incorporated into Phase 2 of the study. Throughout the duration of Phase 2, single genes continue to be added to the study list.

Phases 1 and 2: Copy number variants			
16p11.2 deletions	16p11.2 duplications	1q21.1 deletions	1q21.1 duplications

Phase 2: Genes associated with features of autism				
ACTL6B	CHD2	GRIN2A	MBD5	SETBP1
ADNP	CHD8	GRIN2B	MED13L	SETD5
AHDC1	CSNK2A1	HIVEP2	PACS1	SMARCA4
ANK2	CTBP1	HNRNPH2	PBRM1	SMARCC1
ANKRD11	CTNNB1	KAT6A	POGZ	SMARCC2
ARID1B	CUL3	KATNAL2	PPP2R5D	STXBP1
ASH1L	DDX3X	KDM5B	PTCHD1	SUV420H1
ASXL3	DSCAM	KDM6B	PTEN	SYNGAP1
BAF190	DST	KMT2C	PURA	TBR1
BCL11A	DYRK1A	KMT2E	REST	
CHAMP1	FOXP1	KMT5B	SCN2A	

Phase 2 Exclusion Criteria

Families in Phase 1 of Simons VIP who declined participation in Phase 2 are not included in the Phase 2 data set. Families are required to speak, read and understand English fluently to complete consent and behavioral measures, which are only available in English at this time.

Exclusion criteria for non-carrier control siblings:

- Known genetically-confirmed mutations OTHER THAN the one being studied in the family resulting in effects on neurocognitive outcome.
- Other early environmental insults that may affect neurocognitive outcome, such as fetal alcohol syndrome, severe birth asphyxia, severe prematurity (gestational age < 30 weeks), neurological disease or injury, certain prenatal infections or gestational drug exposure.

Genetic findings are screened for inclusion by genetic counselors. Flags are available to indicate inconsistencies or confounds, such as variants of uncertain significance or environmental insult.

SPARK Participation Criteria

The Simons Foundation Powering Autism Research for Knowledge (SPARK; <https://sparkforautism.org>) aims to recruit, engage and retain a community of 50,000 individuals with ASD and their family members. SPARK families enroll online at www.SPARKforAutism.org, provide saliva samples for genetic analysis and agree to be contacted for future research opportunities.

The phenotypic data currently available includes basic family and medical history information, the Social Communication Questionnaire-Lifetime (SCQ-L), background history information, the Repetitive Behaviors Scale-Revised (RBS-R), and the Developmental Coordination Disorder Questionnaire (DCDQ).

Exome sequencing data from 469 trios (biological parents and child with autism) is currently available. Data from over 4,000 trios is expected to be available in fall 2018.

Inclusion Criteria

Diagnostic information

- All individuals with a professional diagnosis of ASD can participate.
- ASD includes DSM-IV Asperger's syndrome, autism/autistic disorder, Rett Syndrome, Childhood Disintegrative Disorder and pervasive developmental disorder-not otherwise specified (PDD-NOS).

Age

- Individuals of all ages are welcome to participate in SPARK. Children under the age of 12 months registered as ASD are not published until diagnosis can be confirmed or updated with the family.

Family members

- Biological parents and up to one unaffected full biological sibling can participate in SPARK (multiplex families may include multiple unaffected biological siblings). One non-biological guardian may enroll to provide data for participating probands and siblings.

Exclusion Criteria

Residence

- Only residents of the United States can participate in SPARK at this time.

Language

- Participants must be able to read and understand English.

Family members

- Only full biological family members participate in SPARK biospecimen collection.

SSC Available Instruments by Family Role Table

Note: Protocol modifications are sometimes made to accommodate new and/or changed instruments: not all instruments are available for all participants.

Instrument/Activity	Parents	Probands	Unaffected Siblings
Aberrant Behavior Checklist (ABC) Measure of: Aberrant behaviors Format: Questionnaire Administered when: Included in Parent Questionnaire Packet Completed by: Parent Publisher: Slosson		X	
Adult Behavior Checklist for Ages 19 to 59 (ABCL) Measure of: Problem Behavior Format: Questionnaire Administered when: Included in Parent Questionnaire Packet Completed by: Parent Publisher: ASEBA			X
Autism Diagnostic Interview-Revised (ADI-R) Measure of: Parent report of behaviors related to autism phenotype Format: Direct interview Administered when: SSC Research Evaluation Administered by: Examiner who has achieved research levels of ADI-R reliability according to SSC standards Administered to: Parent Publisher: WPS		X	
Autism Diagnostic Observation Schedule (ADOS) Measure of: Observational measure of autism phenotype Format: Direct examiner observation Administered when: SSC Research Evaluation Administered by: Examiner who has achieved research levels of ADOS reliability according to SSC standards Publisher: WPS		X	

Instrument/Activity	Parents	Probands	Unaffected Siblings
Brief family physical and dysmorphology examination Measure of: Physical anomalies Format: Direct physical exam Administered when: Physical exam Administered by: Medical or clinical staff Author: Judith Miles	X	X	X
Broad Autism Phenotype Questionnaire (BAPQ) Measure of: Broader autism phenotype Format: Self-report questionnaire Administered when: Included in Parent Questionnaire Packet Completed by: Mother and father Publisher: Piven	X		
Child Behavior Checklist for ages 6 to 18 years (CBCL) Measure of: Problem behavior Format: Questionnaire Administered when: Included in Parent Questionnaire Packet Completed by: Parent Regarding whom: Proband and siblings, ages 6 to 18 years Publisher: ASEBA		X	X

Instrument/Activity	Parents	Probands	Unaffected Siblings
<p>Cognitive Testing DAS-II (Differential Ability Scales, Second Edition) Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Clinical/Research staff Administered to: Proband ages 4 to 17 years 11months Publisher: Harcourt Assessments</p> <p>Mullen (Mullen Scales of Early Learning, AGS Edition) Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Clinical/Research staff Publisher: Pearson Assessments</p> <p>WASI (Wechsler Abbreviated Scale of Intelligence) Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Clinical/Research staff Publisher: Harcourt Assessments</p> <p>WISC-IV (Wechsler Intelligence Scale for Children, Fourth Edition) Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Clinical/Research staff Publisher: Harcourt Assessments</p>		X	
<p>Developmental Coordination Disorder Questionnaire (DCDQ) Measure of: motor delays Format: Questionnaire Administered when: Parent Questionnaire Packet Completed by: Parents Author: B.N. Wilson</p>		X	
<p>Diagnosis Summary Form Index of: DSM-IV-TR Diagnoses; Diagnostic Certainty; Clinicians Involved in Making Diagnosis; Deviation IQ, Ratio IQ, and Mental Age scores; Information potentially critical to the evaluation (e.g., undiagnosed problems in parents and siblings). Format: Form Administered when: Following SSC Research Evaluation Completed by: Observing senior psychologist/psychiatrist with clinical/research staff</p>		X	
<p>Interviewer's Clinical Impressions (FHI-I) Measure of: Broader phenotype Format: Rating Administered when: Following ADI-R interview at the SSC Research Evaluation Completed by: ADI-R examiner Publisher: IMGSAAC</p>	X		

Instrument/Activity	Parents	Probands	Unaffected Siblings
Comprehensive Test of Phonological Processing Non-word Repetition (CTOPP-NR) Measure of: speech and memory of sounds and non-words Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Clinical/Research staff Administered to: Probands with a verbal mental age of 60 months Publisher: PRO-ED		X	
Parent Substance Use History Form Format: Questionnaire Administered when: Included in Parent Questionnaire Packet Completed by: Both parents Publisher: SSC	X		
Peabody Picture Vocabulary Test, Fourth Edition (PPVT-4) Measure of: Receptive single-word vocabulary Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Clinical/Research staff Publisher: NCS Pearson, Inc.		X	
Purdue Peg Board Measure of: Fine-motor dexterity Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Clinical/Research staff Publisher: Lafayette Instrument Co.		X	
Raven's Standard Progressive Matrices Measure of: Nonverbal problem solving Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Clinical/Research staff Publisher: Harcourt Assessments		X	
Repetitive Behavior Scale-Revised (RBS-R) Measure of: Repetitive behaviors Format: Questionnaire Administered when: Included in Parent Questionnaire Packet Completed by: Parents Publisher: Bodfish		X	
Social Communication Questionnaire (SCQ-L) (Parent report) Measure of: Screen of ASD markers Format: Questionnaire Administered when: Included in Parent Questionnaire Packet Completed by: Parents Regarding whom: Parents report on probands (age 4 years to 17 years 11 months) and siblings (age 4 and up) Publisher: WPS		X	X

Instrument/Activity	Parents	Probands	Unaffected Siblings
<p>Social Communication Questionnaire (SCQ-C) (Teacher report) Measure of: Screen of ASD markers Format: Questionnaire Administered when: Included in Teacher Questionnaire Packet Completed by: Teachers Regarding whom: Teachers report on probands age 4 years to 17 years 11 months and siblings age 4 and up Publisher: WPS</p>		X	X
<p>Social Responsiveness Scale (SRS-ARV) – Adult Research Version Measure of: Autistic traits on a continuous scale Format: Questionnaire Administered When: Included in Parent Questionnaire Packet Completed by: Parents Regarding whom: Mother reports on father; father reports on mother; parent reports on adult sibling ages 19 and up Publisher: WPS</p>	X		
<p>Social Responsiveness Scale (SRS) – Parent report Measure of: Autistic traits on a continuous scale Format: Questionnaire Administered when: Included in Parent Questionnaire Packet Completed by: Parents Regarding whom: Parents report on probands (age 4 years to 17 years, 11 months) and siblings (age 4 to 18 years) Publisher: WPS</p>		X	X
<p>Social Responsiveness Scale (SRS) – Teacher report Measure of: Autistic traits on a continuous scale Format: Questionnaire Administered when: Included in Teacher Questionnaire Packet Completed by: Teachers Regarding whom: Teachers report on siblings (age 4 to 18 years) and probands (age 4 years to 17 years 11 months) Publisher: WPS</p>		X	X
<p>SSC Background History Form Index of: Contact information, family demographic information; parent report of proband's developmental milestones Format: Phone interview Administered when: Included in Parent Questionnaire Packet Completed by: Parent Publisher: SSC</p>	X	X	X

Instrument/Activity	Parents	Probands	Unaffected Siblings
SSC Family Pedigree Form Measure of: Phenotype of ancestry Format: Phone interview Administered when: Parent phone interview following return of Parent Questionnaire Packet Administered by: Clinical/Research staff Administered to: Parent Regarding whom: Immediate and extended family of proband Publisher: SSC		X	
SSC Height, Weight, Head Circumference Form Measure of: Physical characteristics Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Physician or clinical/research staff Publisher: SSC	X	X	X
SSC Medical History Interview Measure of: Family medical history Format: Telephone Interview Administered when: Parent phone Interview following return of Parent Questionnaire Packet Administered by: Clinical/Research staff Publisher: SSC	X	X	X
SSC Temperature Form Measure of: Temperature Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Physician or clinical/research staff Publisher: SSC		X	
SSC Treatment History Form Measure: Past participation in therapeutic treatments Format: Phone interview Administered when: Parent phone interview prior to research evaluation Administered by: Clinical/Research staff Administered to: Parent Publisher: SSC		X	

Instrument/Activity	Parents	Probands	Unaffected Siblings
<p>Teacher Report Form (TRF)</p> <p>Measure of: Problem behavior and school functioning</p> <p>Format: Questionnaire</p> <p>Administered when: Included in Teacher Questionnaire Packet</p> <p>Completed by: Teacher most knowledgeable</p> <p>Regarding whom: Siblings (ages 6 to 18 years) and proband (ages 6 years to 17 years 11 months)</p> <p>Publisher: ASEBA</p> <p>OR: Caregiver-Teacher Report Form (C-TRF)</p> <p>Measure of: Problem behavior</p> <p>Format: Questionnaire</p> <p>Administered when: Included in Teacher Questionnaire Packet</p> <p>Completed by: Teacher/Daycare provider most knowledgeable</p> <p>Regarding: Siblings between 1 year 6 months and 5 years 11 months of age and probands between 4 years and 5 years 11 months</p> <p>Publisher: ASEBA</p>		X	X
<p>Vineland Adaptive Behavior Scale-II (VABS-II)</p> <p>Measure of: Adaptive behavior</p> <p>Format: Interview</p> <p>Administered when: Parent phone interview regarding siblings and parent interview about proband following ADI-R at SSC Research Evaluation</p> <p>Administered by: Clinical/Research staff</p> <p>Administered to: Parents</p> <p>Publisher: Pearson</p>		X	X

SSC@IAN Follow-Up Study Available Instruments by Role Table

Instrument/Activity	Parents	Probands	New Siblings	Unaffected Siblings
Aberrant Behavior Checklist (ABC) Measure of: Aberrant behaviors Format: Online Questionnaire Completed by: Parent Publisher: Slosson		X		
Adult Behavior Checklist for Ages 19 to 59 (ABCL) Measure of: Problem behavior Format: Online Questionnaire Completed by: Parent Regarding whom: Proband and siblings, ages 6 to 18 years Publisher: ASEBA		X		X
Child Behavior Checklist for ages 6 to 18 years (CBCL) Measure of: Problem behavior Format: Online Questionnaire Completed by: Parent Regarding whom: Proband and siblings, ages 6 to 18 years Publisher: ASEBA		X		X
Communication and Symbolic Behavior Scales (CSBS) Measure of: Communication Delays Format: Online Questionnaire Completed by: Parent Regarding whom: New siblings (baby sibs) Publisher: Brookes			X	

Instrument/Activity	Parents	Probands	New Siblings	Unaffected Siblings
Repetitive Behavior Scale-Revised (RBS-R) Measure of: Repetitive behaviors Format: Online Questionnaire Completed by: Parents Publisher: Bodfish		X		
Social Responsiveness Scale 2 (SRS-2) – Parent report Measure of: Autistic traits on a continuous scale Format: Online Questionnaire Completed by: Parents Regarding whom: Probands (age 4 years to 17 years 11 months) and siblings (age 4 to 18 years) Publisher: WPS		X		X
Social Responsiveness Scale 2 (SRS-2) – Adult (relative/other report) Measure of: Autistic traits on a continuous scale Format: Online Questionnaire Completed by: Parents, spouses, or other appointed adults Regarding whom: Parents, independent probands (age >18), siblings (age >18) Publisher: WPS	X	X		X
SSC@IAN Adult Proband Update Form Measure of: Diagnosis, medical and educational updates, employment and living situation Format: Online Questionnaire Completed by: Independent Probands >18, parents Regarding Whom: Proband (>18) Publisher: SSC		X		
SSC@IAN ASD Diagnosis and Medical History Update Form Measure of: Diagnosis and brief medical history updates Format: Online Questionnaire Completed by: Parents Regarding Whom: Proband, designated sibling (<18) Publisher: SSC		X		

Instrument/Activity	Parents	Probands	New Siblings	Unaffected Siblings
SSC@IAN Education History Follow-Up Measure of: Education history Format: Online Questionnaire Completed by: Parent Regarding Whom: Proband, designated sibling (age <18) Publisher: SSC		X		X
SSC@IAN Family Medical History Follow-Up Measure of: Family medical history Format: Online Questionnaire Completed by: Parent Publisher: SSC	X	X	X	X
SSC@IAN Medical and Education History Follow-Up Measure of: Medical and educational history Format: Online Questionnaire Completed by: Parents and Designated Siblings (age >18) Regarding whom: Unaffected Siblings Publisher: SSC				X
SSC@IAN Medications History Measure of: Proband medication history Format: Online Questionnaire Completed by: Parent Publisher: SSC		X		

Simons VIP Available Instruments by Role Table

Simons VIP Phase 1: In-clinic study measures

Phase 1 of this project involved in-person evaluations at clinical study sites. Participants underwent extensive psychological and neurological testing. As a pilot project, Phase 1 also included testing a small number of 1q21.1 deletion and duplication carriers using the identical protocol. Phase 1 ended in early 2014.

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
Neuropsychological Assessments						
Autism Diagnostic Interview-Revised (ADI-R) Measure of: Parent report of behaviors related to autism phenotype (ages up to 25) Format: Direct interview Administered when: Clinical evaluation visit Administered by: Examiner who has achieved research levels of ADI-R reliability according to Simons VIP standards Administered to: Parent Publisher: WPS	X~	X*~	X~	X~		
Autism Diagnostic Observational Schedule (ADOS) Measure of: Observational measure of autism phenotype Format: Direct examiner observation Administered when: Clinical evaluation visit Administered by: Examiner who has achieved research levels of ADOS reliability according to Simons VIP standards Administered to: All carriers and siblings over 30 months of age with a nonverbal mental age of at least 16 months. Also non-carrier parents with a confirmed or suspected diagnosis of autism and/or SRS-ARV T score above 60 Publisher: WPS	X	X*	X	X		
Vineland Adaptive Behavior Scale-II (VABS-II) Measure of: Adaptive behavior Format: Interview Administered when: Clinical evaluation visit, or parent phone interview prior to Parent Questionnaire Packet Administered by: Clinical/Research staff Administered to: Parents Publisher: Pearson	X	X*	X	X		X

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
<p>Autism Diagnostic Observational Schedule for Toddlers (ADOS-T) Measure of: Observational measure of autism phenotype Format: Direct examiner observation Administered when: Clinical evaluation visit Administered by: Examiner who has achieved research levels of ADOS reliability according to Simons VIP standards Administered to: Carriers and siblings less than 30 months of age who are walking, are not yet using simple phrases and have a nonverbal mental age of at least 12 months. Publisher: WPS</p>			X	X		
<p>Cognitive Testing DAS-II (Differential Ability Scales, Second Edition) Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Clinical/Research staff Administered to: Proband ages 4 to 17 years 11 months Publisher: Harcourt Assessments</p> <p>Mullen (Mullen Scales of Early Learning, AGS Edition) Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Clinical/Research staff Publisher: Pearson Assessments</p> <p>WASI (Wechsler Abbreviated Scale of Intelligence) Format: Direct assessment Administered when: SSC Research Evaluation Administered by: Clinical/Research staff Publisher: Harcourt Assessments</p>	X	X	X	X	X	X
<p>Delis-Kaplan Executive Function System (DKEFS) Measure of: Executive function Format: Direct assessment Administered when: fMRI/MEG visit Administered by: Clinical staff Administered to: Carriers participating in fMRI/MEG and all controls, ages 8 to 89 years Publisher: Pearson Assessments</p>	X		X		X	X
<p>Purdue Pegboard Measure of: Fine motor dexterity Format: Direct assessment Administered when: Clinical evaluation visit Administered by: Clinical or research staff Administered to: All family members aged 4 years and up Publisher: Lafayette Instruments</p>	X	X	X	X	X	X

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
<p>Movement Assessment Battery for Children-Second Edition (Movement ABC-2)</p> <p>Measure of: Motor function Format: Direct assessment Administered when: Clinical evaluation visit Completed by: Clinical or research staff Administered to: Child carriers, ages 3 years to 16 years 11 months Publisher: Pearson Assessments</p>			X			
<p>Clinical Evaluation of Language Fundamentals (CELF-4)</p> <p>Measure of: Expressive and receptive language ability Format: Direct assessment Administered when: Clinical evaluation visit Administered by: Clinical staff Administered to: Carriers participating in fMRI/MEG, and all controls, ages 5 to 21 years Publisher: Pearson Assessments</p> <p><i>Note:</i> o Ages 5-8: word structure, recalling sentences, concepts and directions, and formulated sentences. o Ages 9-12: recalling sentences, concepts and directions, formulated sentences and word classes. o Ages 13 and up: recalling sentences, formulated sentences, word classes and word definitions.</p>	X#		X#		X	X
<p>CASL (Comprehensive Assessment of Spoken Language)</p> <ul style="list-style-type: none"> • Measure Of: Spoken language • Format: Direct assessment • Administered when: In-person visit • Administered by: Clinical/Research staff • Administered to: Probands, siblings, ages 3 years to 21 years <p>Publisher: Pearson Assessments</p>			X	X		
<p>CCC-2 (Children's Communication Checklist-2)</p> <p>Measure of: Language and Pragmatic Skills Format: Questionnaire Administered When: Questionnaire Packet, prior to in-person visit Completed by: Parent Regarding whom: Proband, siblings and controls, ages 4 years to 16 years 11 months who speak in full sentences Publisher: Pearson Assessments</p>			X	X		X

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
<p>Comprehensive Test of Phonological Processing – Non-Word Repetition task (CTOPP)</p> <p>Measure of: Phonological processing Format: Direct assessment Administered when: Clinical evaluation visit Administered by: Clinical or research staff Administered to: Carriers, siblings, parents and controls (ages 5 to 24 years) Publisher: Pro Ed Inc.</p>	X	X	X	X	X	X
<p>MacArthur Communicative Development Inventory – Words and Gestures, and Words and Phrases versions (MCDI)</p> <p>Measure of: Early language use Format: Questionnaire Administered when: Questionnaire Packet, prior to clinical evaluation visit Completed by: Parents Regarding whom: Carriers, siblings and controls, ages 8 to 30 months or pre-phrase speech Publisher: Pearson Assessments</p>			X	X		
<p>WIAT III (Wechsler Individual Achievement Test, Version III)</p> <p>Measure of: Academic achievement Format: Direct assessment Administered when: Clinical evaluation visit Administered by: Clinical or research staff Administered to: All carriers (in grade 1 up through age 50 years 11 months) Publisher: Pearson Assessments <i>Note: Only sections on reading comprehension, word reading, numerical operations, sentence composition (which includes sentence building and sentence completion) were completed.</i></p>	X		X		X	X
Behavioral Inventories						
<p>Adult Behavior Checklist (ABCL)</p> <p>Measure of: Problem behavior Format: Questionnaire Administered when: Questionnaire packet, prior to clinical evaluation visit Completed by: Participant's parent (or other well-known adult, e.g., spouse or other relative) Regarding: Adult carriers, adult controls and non-parent adults, ages 19 to 59 years Publisher: ASEBA</p>	X	X*			X	

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
<p>Child Behavior Checklist for ages 6 to 18 years (CBCL) Measure of: Problem behavior Format: Questionnaire Administered when: Questionnaire Packet, prior to clinical evaluation visit Completed by: Parent Regarding whom: Carriers, siblings and controls, ages 6 to 18 years Publisher: ASEBA</p>			X	X		X
<p>Diagnostic Interview Schedule for Children (DISC) – Young Child and Child versions Measure of: Psychiatric disorders Format: Direct assessment or phone interview Administered when: Clinical evaluation visit or phone interview Administered by: Clinical or research staff Administered to: Parent (no child self-report, only parent report on child) Regarding whom: Carriers, siblings and controls, ages 3 years to 5 years 11 months (young child version) or 6 to 17 years (regular child version) Publisher: Columbia University, Casia Software</p>			X	X		X
<p>Symptom Checklist-90-Revised (SCL-90-R) Measure of: Psychiatric disorders Format: Questionnaire Administered when: Questionnaire Packet, prior to clinical evaluation visit Completed by: Self Regarding whom: Parent, adult carrier, adult siblings and controls (ages 18 years and up) Publisher: Pearson Assessments</p>	X	X			X	
<p>Broader Autism Phenotype Questionnaire (BAPQ) Measure of: Broader autism phenotype Format: Self-report questionnaire Administered when: Questionnaire Packet, prior to clinical evaluation visit Completed by: Parents, adult siblings, adult non-parent/non-sibling carriers and adult controls Regarding whom: Self Publisher: Piven</p>	X	X			X	

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
<p>Behavior and Stereotyped Interest Questionnaire (BSIQ) Measure of: Repetitive behavior Format: Direct interview Administered when: Clinical evaluation visit Administered to: Parent Administered by: A clinician who is reliable on the ADI (or trained by Simons VIP staff to administer this measure, if a site does not administer ADI) Regarding whom: Child carriers, child siblings and child controls Developed by: Harvard University</p>			X	X		X
<p>Parenting Stress Index (PSI) Measure of: Parental stress Format: Questionnaire Administered when: Questionnaire Packet, prior to clinical evaluation visit Completed by: Each parent, regarding children currently under 18 years Regarding whom: children under 18 Publisher: WPS</p>			X	X		
<p>SCQ (Social Communication Questionnaire—Lifetime) Measure of: Screen of ASD markers Format: Questionnaire Administered when: Questionnaire Packet, prior to in-person visit Completed by: Parent Regarding whom: Proband, siblings and controls ages 4 years to 17 years 11 months, with a mental age over 2 years Publisher: WPS</p>			X	X		X
<p>Social Responsiveness Scale (SRS) Measure of: Autistic traits Format: Questionnaire Administered when: Questionnaire Packet, prior to clinical evaluation visit Completed by: Parent Regarding whom: Carriers, siblings and controls, ages 4 years to 18 years 11 months Publisher: WPS</p>			X	X		X
<p>Social Responsiveness Scale-Adult Research Version (SRS-ARV) Measure of: Autistic traits Format: Questionnaire Administered when: Questionnaire Packet, prior to clinical evaluation visit Completed by: Parents, adult carriers, adult siblings and adult controls Regarding whom: Mother reports on father; father reports on mother; parent reports on adult carriers and siblings ages 19 years and up Publisher: WPS</p>	X	X			X	

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
Other Measures						
Dysmorphology—3dMD Pictures Measure of: Broader autism phenotype Format: Record form, 3-dimensional (3D) rendered photograph file Administered when: Clinical evaluation visit Completed by: Research staff	X	X	X	X		
Education History Interview Measure of: Education history Format: Interview Administered when: Clinical evaluation visit Administered by: Clinical or research staff Administered to: Parent (for child carrier) or self (for adult carrier)	X		X			
Family Structure Form (Pedigree) Measure of: Family membership and structure, medical disorders per Medical History Interview Format: Phone interview Administered when: Parent phone interview prior to Parent Questionnaire Packet Administered by: Clinical or research staff Administered to: Parent Regarding whom: Proband's immediate and extended family	X		X			
Intervention History Interview Measure of: Intervention history Format: Interview Administered when: Clinical evaluation visit Administered by: Clinical or research staff Administered to: Parent (for child carrier) or self (for adult carrier) Regarding whom: Carriers	X		X			
Medical History Interview (MHI – Adult and Child versions) Measure of: Family medical history Format: Telephone interview Administered when: Parent phone interview prior to Parent Questionnaire Packet Administered by: Clinical/Research staff Administered to: Mother Regarding whom: Proband's immediate and extended family Publisher: N/A	X		X			

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
Medication Interview Measure of: Medication use Format: Interview Administered when: Parent phone interview prior to Parent Questionnaire Packet Administered by: Clinical/Research staff Administered to: Parent, or self (for adult proband) Regarding whom: Proband Publisher: N/A	X		X			
Mother Other Pregnancy Questionnaire Measure of: Pregnancy history — non-live births Format: Questionnaire Administered when: Questionnaire Packet, prior to in-person visit Completed by: Parent Regarding whom: Biological mothers of probands Publisher: N/A	X	X				
Previous Diagnosis Interview (Parent report on child, Self-Report Versions) Measure of: Previous diagnoses (additional information) Format: Interview Administered when: In-person visit Administered by: Clinical staff Administered to: Parents/Self Regarding whom: Child probands/adult probands Publisher: N/A	X	X				
Puberty Questionnaire (versions for boys, girls, men, women) Measure of: Puberty status Format: Questionnaire Administered when: Questionnaire Packet, prior to in-person visit Completed by: Parent of proband, adult proband Regarding whom: All probands Publisher: N/A	X	X	X			
Simons VIP Background History Form Index of: Family demographic information, parent report of carrier's developmental milestones Format: Questionnaire Administered when: Questionnaire Packet, prior to clinical evaluation visit Completed by: Parent Regarding whom: Family and carriers and child controls Publisher: Simons VIP	X		X		X	X

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
<p>Simons VIP Feeding Questionnaire Measure of: Eating behavior Format: Questionnaire Administered when: Questionnaire Packet, prior to clinical evaluation visit Completed by: Parent Child Form: Carriers, siblings and controls with cognitive age of more than 24 months to 12 years Adolescent Form: Carriers, siblings and controls, ages 13 years to 17 years 11 months</p>			X	X		X
<p>Eating in Absence of Hunger — Adult Self-Report, Parent Report on Child, and Child Self-Report versions (EAH) Measure of: Eating behavior Format: Questionnaire Administered when: Questionnaire Packet, prior to clinical evaluation visit Completed by: Parents Regarding whom: Adults about themselves; parents about carriers, siblings and controls; children about themselves as able</p>	X	X	X	X	X	X
<p>LOC (Loss of Control) — Parent report on child, Self-report versions Measure of: Eating behavior Format: Questionnaire Administered when: Questionnaire Packet, prior to in-person visit Completed by: Self if possible, or parent about subject (see note) Regarding whom: Parents, siblings, controls and proband with cognitive age 60m+ Publisher: N/A Note: Depending on cognitive age of child, the child may self-report. If a child self-report is obtained, a parent report on that child should also be obtained.</p>	X	X	X	X	X	X
<p>Pediatric Sleep Questionnaire Measure of: Sleep habits Format: Questionnaire Administered when: Questionnaire Packet, prior to in-person visit Completed by: Parent Regarding whom: Child probands, child siblings, child controls Publisher: N/A, based on a sleep questionnaire by Ron Chervin, M.D.</p>			X	X		X

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
Adult Sleep Questionnaire Measure of: Sleep habits Format: Questionnaire Administered when: Questionnaire Packet, prior to in-person visit Completed by: Parents regarding self, adult probands, adult siblings, adult controls Regarding whom: self Publisher: N/A	X	X			X	
Edinburgh Handedness Inventory <ul style="list-style-type: none"> • Measure of: Handedness • Format: Questionnaire • Administered when: In-person visit • Administered by: Clinical/Research staff • Administered to: Everyone ages > 2 years, including controls, use self-report 6 and up when able. Publisher: Psychology Press	X	X	X	X	X	X
HWHC Form (Height, Weight, Head Circumference Form) <ul style="list-style-type: none"> • Measure of: Physical characteristics • Format: Direct assessment • Administered when: In-person visit • Administered by: Physician or clinical/research staff • Administered to: Mother, father, proband, siblings and controls Publisher: N/A	X	X	X	X	X	X
Neurological Exam <ul style="list-style-type: none"> • Measure of: Cranial nerve and motor functioning, physician screen of speech production and quality, spine and skin exam • Format: Direct assessment • Administered when: In-person visit • Administered by: Neurologist • Regarding whom: Probands, controls (all ages) • Publisher: N/A 	X	X			X	X

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier	Adult Control	Child Control
Neurologic Record Review Form (NRRF) <ul style="list-style-type: none"> • Measure of: Medical history of neurological conditions • Format: Record form • Completed when: At/After visit • Completed by: SVIP neurologist • Regarding whom: Probands • Publisher: N/A 	X	X				
Structural MRI Scan Results Form Measure of: Primary results from review of structural MRI scans (both preexisting, if acceptable, and scan as part of the study) Format: Record form Completed when: Post-scanning and review of structural scans Completed by: Central imaging staff Regarding whom: Everyone in family with a structural MRI scan	X	X*	X	X		

KEY:

* = Only if adult sibling

~ = If autism spectrum disorder suspected

= Only participants that go on to the functional imaging stage

Simons VIP Phase 2: Online study measures

Phase 2 involves the collection of family medical, developmental and behavioral information through online surveys and phone interviews with families. All families will 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.

Note: Protocol modifications are sometimes made to accommodate new and/or changed instruments; not all instruments are available for all participants.

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier
Interview				
Medical History Interview (MHI – Adult and Child version) Measure of: Family medical history Format: Telephone interview Administered by: Genetic counselor Administered to: Mother or self for adult proband Regarding whom: Proband Publisher: N/A	X		X	
Medication Interview Measure of: Medication use Format: Phone interview Administered by: Genetic counselor/clinical/research staff Administered to: Parent, or self (for adult proband) Regarding whom: Proband	X		X	
Mother Pregnancy Questionnaire Measure of: Pregnancy history Format: Phone survey Administered by: Genetic counselor Completed by: Parent Regarding whom: Biological mothers of probands Publisher: N/A				
Mother Other Pregnancy Questionnaire Measure of: Pregnancy history — non-live births Format: Phone survey Administered by: Genetic counselor Completed by: Mother Regarding whom: Biological mothers of probands Publisher: N/A				

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier
<p>Previous Diagnosis Interview Measure of: Previous diagnoses (additional information) Format: Interview Administered by: Genetic counselor/clinical staff Administered to: Parents or self for adult Regarding whom: Child probands/adult probands, siblings Publisher: N/A</p>	X	X	X	X
<p>Puberty Questionnaire (versions for boys, girls, men, women) Measure of: Puberty status Format: Telephone survey Administered by: Genetic counselor Completed by: Parent of proband, or self for adult proband Regarding whom: All probands Publisher: N/A</p>	X		X	
<p>Vineland Adaptive Behavior Scale-II (VABS-II) Measure of: Adaptive behavior Format: Interview Administered when: parent phone interview Administered by: Clinical/Research staff Administered to: Parents Publisher: Pearson</p>	X	X*	X	X
Online				
<p>Adult Behavior Checklist (ABCL) Measure of: Problem behavior Format: Online Questionnaire Completed by: Participant's parent (or other well-known adult, e.g., spouse or other relative) Regarding: Adult carriers, adult controls and non-parent adults, ages 19 to 59 years Publisher: ASEBA</p>	X	X*		
<p>ARI (Adult Routines Inventory) Measure of: Repetitive behavior Format: Online Questionnaire Completed by: Parent/Adult Regarding whom: Self Developed by: D. Evans</p>	X	X		
<p>Behavior and Stereotyped Interest Questionnaire (BSIQ; Online Version) Measure of: Repetitive behavior Format: Online Questionnaire Completed by: Parent Regarding whom: Child carriers, child siblings Developed by: Ellen Hanson, Harvard University</p>			X	X

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier
Broader Autism Phenotype Questionnaire (BAPQ) Measure of: Broader autism phenotype Format: Online Questionnaire Completed by: Parents, adult siblings, adult non-parent/non-sibling carriers and adult controls Regarding whom: Self Publisher: Piven	X	X		
Child Behavior Checklist for ages 6 to 18 years (CBCL) Measure of: Problem behavior Format: Online Questionnaire Completed by: Parent Regarding whom: Carriers, siblings and controls, ages 6 to 18 years Publisher: ASEBA			X	X
Childhood Routines Inventory-Revised (CRI) Measure of: Repetitive behavior Format: Online Questionnaire Completed by: Parent/Guardian Regarding whom: Probands and siblings Developed by: D. Evans			X	X
GO-LIFE (Adult) Measure of: Unusual perceptual experiences Format: Online Questionnaire Completed by: Parent/Adult Regarding whom: self Developed by: D. Evans	X	X		
GO-LIFE – C Measure of: Unusual perceptual experiences Format: Online Questionnaire Completed by: Parent/Guardian Regarding whom: Probands and siblings Developed by: D. Evans	X			
Modified Checklist for Autism in Toddlers (MCHAT) Measure of: Screening for early risk of ASD Format: Online Questionnaire Completed by: Parent Regarding whom: Probands and siblings age 16 to 30 months Author: Robins, Fein & Barton (1999).			X	X
Simons VIP Background History Form Index of: Contact information, family demographic information, parent report of carrier's developmental milestones Format: Online Questionnaire Completed by: Parent or self for adult proband Regarding whom: Family and carriers and child controls Publisher: SVIP	X		X	
Social Communication Questionnaire—Lifetime (SCQ) Measure of: Screen of ASD markers Format: Online Questionnaire Completed by: Parent Regarding whom: Proband, siblings and controls ages 4 years to 17 years 11 months Publisher: WPS				X

Full Name	Adult Carrier	Adult Non-carrier	Child Carrier	Child Non-carrier
Social Responsiveness Scale (SRS-2) (School age 4-18; Adult 19+) Measure of: Autistic traits Format: Online Questionnaire Completed by: Parent, or spouse or other relative for adults Regarding whom: Carriers, siblings and controls, ages 4 years to 18 years 11 months; 19+ Publisher: WPS	X	X	X	X
Seizure Survey- Online version Measure of: Self-reported seizure history Format: online questionnaire Completed by: parent/self Regarding whom: carriers Publisher: N/A	X		X	
Teacher Rating Form (TRF- 1.5-5, 6-18) Measure of: Problem behavior Format: Online Questionnaire Completed by: Teacher RegardingWhom: Child proband and non-carrier sibling Publisher: ASEBA			X	X
Teacher SRS-2 Measure of: Autistic traits Format: Online Questionnaire Completed by: Teacher Regarding whom: Child proband and non-carrier siblings, ages 2 years 6 months to 18 years Publisher: WPS			X	X

* Only if adult sibling

SPARK Available Instruments by Family Role Table

Instrument/Activity	Parents	Individuals with ASD	Unaffected Siblings
Basic Medical Screening Questionnaire Measure of: Family and medical history Format: Questionnaire Administered when: Online registration complete Completed by: Parent Publisher: N/A	X	X	X
Social Communication Questionnaire (SCQ) – Lifetime Measure of: Screen of ASD markers Format: Online Questionnaire Completed by: Parents Regarding whom: Probands and siblings age 2 to 17 years 11 months Publisher: WPS		X	X
Repetitive Behaviors Scale – Revised Measure of: Repetitive behaviors Format: Online Questionnaire Completed by: Parent/guardian Regarding whom: Dependent ASD individuals age 3 years and up Publisher: Bodfish		X	
Developmental Coordination Disorder Questionnaire Measure of: Motor delays Format: Online Questionnaire Completed by: Parent/guardian Regarding whom: Dependent ASD individuals age 5-15 years Publisher: B.N. Wilson		X	
Background History Questionnaire – Child Measure of: Demographics and developmental history Format: Online Questionnaire Completed by: Parent/guardian Regarding whom: Dependent ASD individuals Publisher: SFARI		X	
Background History Questionnaire – Sibling Measure of: Demographics and developmental history Format: Online Questionnaire Completed by: Parent/guardian Regarding whom: Non-ASD siblings under 18 years Publisher: SFARI			X
Background History Questionnaire – Adult Measure of: Demographics and developmental history Format: Online Questionnaire Completed by: Parent/guardian Regarding whom: Independent ASD adults over 18 years Publisher: SFARI		X	

Prerequisites for Access to SFARI Base

Required Institutional Documents

Please note that each principal investigator (PI) who wishes to conduct one or more research projects using SFARI data or materials must obtain the following from their institution before applying:

- An electronically signed [Researcher Distribution Agreement \(eRDA\)](#) — The RDA specifies the legal issues pertaining to research data and specimens, and requires approval from the PI's institution. Each PI and collaborator must sign the joinder agreement to the eRDA. Instructions for signing the eRDA will be emailed to the institutional officer directly from collections@sfari.org. (For researchers at for-profit institutions, please email collections@sfari.org to get the modified, corporate RDA.)
- A letter from an institutional review board (IRB) — Each SFARI project led by a PI must be governed by a protocol approved or exempted by an IRB. A protocol specifies the regulatory issues pertaining to the research, and may include co-investigators at multiple institutions. You will need to obtain a PDF copy of the letter from your IRB approving or exempting your protocol.

Online SFARI Application Instructions

1. Create a SF I.D. at <https://id.simonsfoundation.org>.
2. After creating a SF I.D., sign in to SFARI Base (<https://base.sfari.org/ordering>).
3. Affiliate with an institution. If your institution is not already on the list, email collections@sfari.org to request that your institution be added.

In this email, you must include institution name and the names and email addresses for the PI and institutional officer in charge of contracts and agreements.

4. Once the eRDA and joinder agreements are signed, you will be able to submit an application for phenotype data, genetic data, imaging data, biospecimens or to recontact the SSC families.

Simons Simplex Collection Sample Consent Form

http://simonsfoundation.s3.amazonaws.com/share/Policies_and_forms/2014/ssc/UMACC_Adult_Consent_03-2011.pdf

Simons VIP Sample Consent Form

Phase 1

http://simonsfoundation.s3.amazonaws.com/share/Policies_and_forms/2014/svip/SVIPSampleConsent9-13-12.pdf

Phase 2

http://simonsfoundation.s3.amazonaws.com/share/simonsvip/SVIP_Phase_2_Sample_Consent.pdf

SPARK Sample Consent Form

http://simonsfoundation.s3.amazonaws.com/share/Policies_and_forms/2017/SPARK_Sample_Consent_Form.pdf

If you have additional questions, email collections@sfari.org.