SIMONS FOUNDATION

SFARI Base
Simons Searchlight

Researcher Welcome Packet

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Simons Searchlight Phase 2 (2013-current)

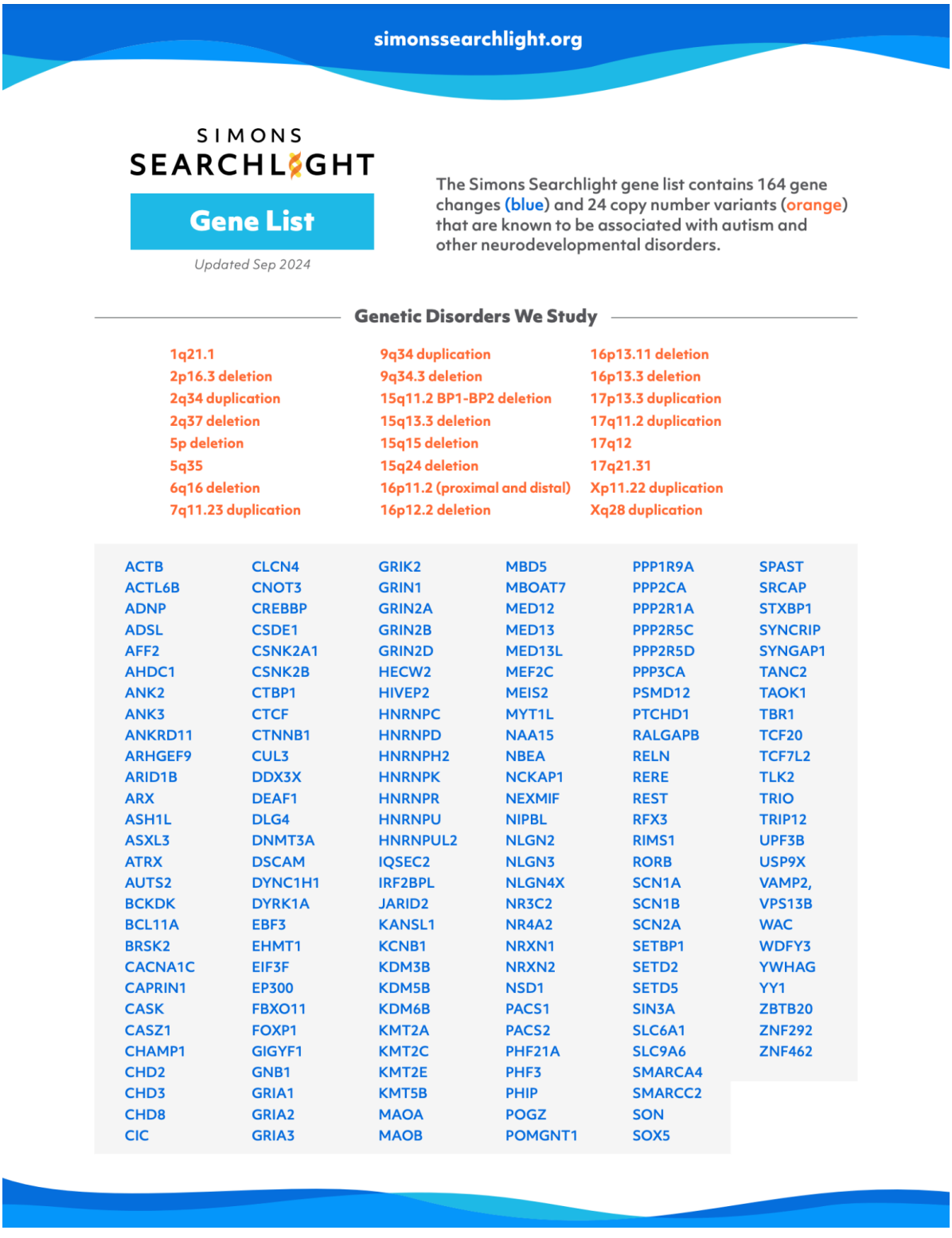
Phase 2 Inclusion Criteria

All participants of Simons Searchlight Phase 1 were invited to participate in Phase 2, which was an online extension of the Phase 1 study. Some Phase 1 families, but not all, consented to participate in Phase 2. Phase 2 participants received a slightly different battery of measures. To see more information about Phase 1, please see pages 4-5.

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, over 180 genes and CNVs associated with neurodevelopmental disabilities and features of autism are eligible to participate in Phase 2 of the study. Throughout the duration of Phase 2, genetic changes continue to be added to the study list.

Carriers in Phase 2 are not excluded on the basis of additional, deleterious genetic mutations or CNVs, nor CNVs that are larger in size than the previously indicated interval, nor distal deletions or duplications in the case of 16p11.2. These cases will be labeled and flagged in the datasets.

Simons Searchlight Gene List



(The most current gene list can be found here.)

Phase 2 Exclusion Criteria

In the beginning of Phase 2, families were required to have fluency in English to complete consent and surveys. In 2020, Searchlight began enrolling families who speak Spanish collecting demographic and medical history. Searchlight is now working to enroll families who speak French, Italian and Dutch. These families' data will be available at a later date.

Genetic findings are screened for inclusion by genetic counselors and non-conforming variants may be excluded from data release on a case by case basis. Flags also are available in the dataset to indicate inconsistencies or confounds in other cases, such as variants of uncertain significance or environmental insult.

Exclusion criteria for non-carrier control siblings:

- Known genetically-confirmed mutations OTHER THAN those being studied in Simons Searchlight and which have neurocognitive effects
- Other early environmental insults that may affect neurocognitive outcome, such as fetal alcohol syndrome, severe birth asphyxia, severe prematurity (gestational age less than 30 weeks), neurological disease or injury, certain prenatal infections or gestational drug exposure
- Half-siblings

Simons Searchlight Phase 2: 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 if individuals are eligible according to age and other guidelines. 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.

A limited number of SCN2A carriers received in-clinic assessments in 2016; measures for this protocol are appended below.

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

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Interview			
Medical History Interview (MHI — Adult and Child version)	Phone interview conducted by a licensed genetic counselor from 2013-2022, parent or self-report	Medical history	Adult carriers Child carriers (discontinued in 2023; replaced with online Medical History Survey - see below)
Medications Interview	Phone interview conducted by a licensed genetic counselor from 2013-2022, parent or self-report	Medication history	Adult carriers Child carriers (discontinued in 2023; replaced with online Medical History Survey - see below)
Pregnancy Questionnaire	Phone interview conducted by a licensed genetic counselor from 2013-2022, self-report	Mother's pregnancy with proband, prenatal exposures	Dependent adult carriers Child carriers (discontinued in 2023; partially replaced with online Medical History Survey - see below)
Other Pregnancy Questionnaire	Phone interview conducted by a licensed genetic counselor from 2013-2022, self-report	Mother's history of pregnancies – not including the proband	Mothers (carrier & non-carrier)

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Previous Diagnosis Interview	Phone interview conducted by a licensed genetic counselor from 2013-2022, parent or self-report	All psychiatric diagnoses	Adult carriers Child carriers (discontinued in 2023; replaced with online Medical History Survey - see below)
Puberty Questionnaire (versions for boys, girls, men, women)	Phone interview conducted by a licensed genetic counselor from 2013-2022, parent or self-report	Puberty status	Adult carriers Child carriers (discontinued in 2023; replaced with online Medical History Survey - see below)
Vineland Adaptive Behavior Scale-II (VABS-II) Pearson	Phone interview conducted by a licensed genetic counselor from 2013-2022, parent report	Adaptive behavior	Adult carriers Child carriers (2013-2021) Child non-carriers (2013-2017)
Online			
Adult Behavior Checklist (ABCL) ASEBA	Online questionnaire, spouse, parent, or trusted adult report	Problem behavior	Dependent adult carriers Ages 19-59
Adult Routines Inventory (ARI) D. Evans	Online, Self-report	Repetitive Behavior	Adult carriers Adult non-carriers (Collected 2013-2015)
Brief Developmental Update SFARI	Online, parent-report	Attainment of walking and speech milestones; means of mobility and communication	Child carriers (implemented 2020)
Behavior and Stereotyped Interests Questionnaire (BSIQ; Online Version) Ellen Hanson, Harvard University	Online, parent-report	Repetitive Behavior	Child carrier Child non-carrier (2013-2016)

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Broader Autism Phenotype Questionnaire (BAPQ) Piven	Online, self-report	Broader autism phenotype	Parents & adult sibling carriers & non-carriers (2013-2015)
Child Behavior Checklist for ages 1.5 to 5 years (CBCL) ASEBA	Online, parent-report	Problem behavior	Child carriers Child non-carriers Ages 1.5-5
Child Behavior Checklist for ages 6 to 18 years (CBCL) ASEBA	Online, parent-report	Problem behavior	Child carriers Child non-carriers Ages 6-18* *Individuals must be ambulatory/verbal
Childhood Routines Inventory-Revised (CRI-R) D. Evans	Online, parent-report	Repetitive behavior	Child carriers Child non-carriers (until 2015)
Children's Sleep Habits Questionnaire (CSHQ)	Online, parent report	Assessment for sleep disorders	Child Carriers Dependent adult carriers (implemented 2021)
GO-LIFE (Adult) D. Evans	Online, self-report	Unusual perceptual experiences	Adult non-carriers Adult carriers (until 2015)
GO-LIFE (Child) D. Evans	Online, parent-report	Unusual perceptual experiences	Child carriers Child non-carriers (until 2015)
Medical History Survey (Online, Baseline & Longitudinal Versions) SFARI	Online, parent-report (2022-present)	Medical history; medication history; mother's pregnancy with proband; psychiatric diagnoses; puberty status	Adult carriers Child carriers
Modified Checklist for Autism in Toddlers (MCHAT) Robins, Fein & Barton (1999)	Online, parent-report	Screening for early risk of ASD	Child carriers Child non-carriers Ages 16-30 months (until 2018)

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Background History Form SFARI	Online, parent-report	Contact information, family demographic information, parent report of carrier's developmental milestones	Adult carriers Child Carriers
Learning to Read SFARI	Online, parent-report	Captures whether a child has learned to read, and what learning system was used	Child/dependent carriers (discontinued 2021)
Qi-Disability (Jenny Downs)	Online, parent-report	Quality of life for children and teens with intellectual disability	Child and adult dependent carriers
Skin & Dental Survey	Online, parent-report	Information on teeth eruption, dental & skin health	Child carriers Adult carriers Non-carrier siblings (discontinued 2021)
Sleep Related Breathing Disorder Questionnaire (SRBD) Ronald Cervin	Online, parent report	Symptoms related to snoring, sleepiness, and inattention & hyperactivity	Child carriers Dependent adult carriers
Sleep Supplement Survey SFARI	Online, parent report	Questions of sleep habits and schedules	Child carriers Dependent adult carriers
Social Communication Questionnaire—Lifetime (SCQ) WPS	Online, parent-report	Screen of ASD markers	Child carriers Child non-carriers Ages 4 – 19 years, 11mo (non-carrier sibling administration was discontinued in 2015)

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Social Responsiveness Scale (SRS-2) WPS	Online, parent or spouse/trusted adult	Autistic traits	Child carriers* Child non-carriers* Dependent adult carriers* Non-carrier parents (Individuals must be ambulatory/verbal. In 2013-2020 all genetic conditions; starting 2021, CNVs only; parents d/c in 2018) School age administration ages 4-18y/11m (assigned if Vineland-3 Expressive Language age equivalent >=3); adult administration 19+
Seizure Survey- Online version I SFARI	Online, parent-report	Seizure history	Child carriers Adult carriers (2015-2020)
Seizure Survey- Online version II with Annual Update (Longitudinal) SFARI	Online, parent-report	Seizure history	Child carriers Adult carriers (implemented 2021)
Skin and Dental Survey SFARI	Online, parent/self-report	Ectodermal conditions	Adult carriers Child carriers Child non-carriers (2018-2021)
Teacher Rating Form (TRF- 1.5-5, 6-18) ASEBA	Online, teacher-report	Problem behavior	Child Carriers Child non-carriers (2013-2015)
Teacher SRS-2 WPS	Online, teacher-report	Autistic traits	Child carriers Child non-carriers (2013-2015)
Vineland Adaptive Behavior Scale-3 (VABS-3) Pearson	Online, parent report	Adaptive behavior	Child and dependent adult carriers (implemented 2021)

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Additional measures *only for SCN2A Clinic Protocol,* Collected in 2016			
Autism Diagnostic Interview-Revised (ADIR) WPS	In-clinic administration by an Examiner who has achieved research levels of ADI-R reliability according to SFARI standards, parent-report	Behaviors related to autism phenotype	Adult carriers Children carriers Ages 2-25, only if ASD was suspected
Autism Diagnostic Observation Schedule (ADOS) WPS	In-clinic administration, by an Examiner who has achieved research levels of ADOS reliability according to SFARI standards, direct examiner observation	Observational measure of autism phenotype	Child Carriers* Child non-carriers* *over 30 months of age with a nonverbal mental age of at least 16 months. Adult non-carrier parents with a confirmed or suspected diagnosis of ASD and/or SRS-ARV t-score above 60
Cognitive Testing Differential Ability Scales Version II (DAS-II) Harcourt Assessments Mullen Scales of Early Learning, AGS Edition (Mullen) Pearson Assessments Wechsler Abbreviated Scale of Intelligence (WASI) Harcourt Assessments	In-clinic administration, direct examiner assessment	Intellectual ability	Adult carriers Child carriers

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
CASL (Comprehensive Assessment of Spoken Language) Pearson	In-clinic administration, direct assessment	Spoken language	Carriers Non-carrier siblings Aged 3-21
Education History Interview SFARI	In-clinic interview, parent/self-report	Education history	Adult carriers Child carriers
Intervention History Interview SFARI	In-clinic interview, parent/self-report	Intervention history	Adult carriers Child carriers
Height, Weight, Head Circumference Form (HWHC) SFARI	Direct assessment	Physical characteristics	Child carriers Adult carriers
Repetitive Behavior Scale- Revised (RBSR) Bodfish	In-clinic interview, parent/guardian report	Repetitive behaviors	Carriers, 3 years and older

Simons Searchlight (Simons VIP) Phase 1 (2010-2014)

Simons Searchlight is a SFARI initiative that aims to better understand genetic neurodevelopmental conditions, specifically those associated with autism spectrum disorder (ASD). Previously named Simons Variation in Individuals Project (Simons VIP), the name change aims to better reflect the program's mission to shed light on these conditions by building strong partnerships between researchers and families.

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. Simons Searchlight (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, as well as 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).

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.

Non-familial 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 non-familial controls

- 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
- in non-familial controls: dysmorphic features
- in non-familial controls: genetic findings (blood draw and subsequent testing)
- in non-familial controls: history of psychiatric or developmental problems per behavior ratings or observation (including significant substance abuse) that may confound interpretation of comparisons, or family history of autism or other developmental disorder.

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)

Simons Searchlight Phase 1: In-clinic study measures (collected prior to 2015)

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.

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Neuropsychological Assessments			
Autism Diagnostic Interview-Revised (ADIR) WPS	In-clinic administration by an Examiner who has achieved research levels of ADI-R reliability according to SFARI standards, parent interview	Behaviors related to autism phenotype	Adult carriers* up to 25 years old Children carriers* Adult and children Non-carrier siblings* up to 25 years old *if ASD is suspected
Autism Diagnostic Observation Schedule (ADOS) WPS	In-clinic administration, by an Examiner who has achieved research levels of ADOS reliability according to SFARI standards, direct examiner observation	Observational measure of autism phenotype	Adult carriers Adult non-carrier siblings Child carriers Child non-carriers
Vineland Adaptive Behavior Scale-II (VABS-II) Pearson	In-clinic parent interview, or parent phone interview	Adaptive behavior	Adult carriers Adult non-carrier siblings Child carriers Child non-carriers Non-familial child controls

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Autism Diagnostic Observation Schedule for Toddlers (ADOS-T) WPS	In-clinic administration, by an Examiner who has achieved research levels of ADOS reliability according to SFARI standards, direct examiner observation		Carriers and siblings less than 30 months of age who were walking, not yet using simple phrases and had a nonverbal mental age of at least 12 months.
Cognitive Testing DAS-II (Differential Ability Scales, Second Edition) Harcourt Assessments Mullen (Mullen Scales of Early Learning, AGS Edition) Pearson WASI (Wechsler Abbreviated Scale of Intelligence) Harcourt Assessments	In-clinic administration, direct assessment	Intellectual ability	Adult carriers Adult non-carriers Child carriers Child non-carriers Non-familial adult and child controls
Delis-Kaplan Executive Function System (DKEFS) Pearson	In-clinic administration, direct assessment	Executive functioning	Carriers participating in fMRI/MEG and all controls aged 8 to 89 years Non-familial adult and child controls
Purdue Pegboard Lafayette Instruments	In-clinic administration, direct assessment	Fine motor dexterity	All Phase 1 participants aged 4 years and older Non-familial adult and child controls

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Movement Assessment Battery for Children-Second Edition (Movement ABC-2) Pearson	In-clinic administration, direct assessment	Motor function	Child carriers aged 3 years to 16 years 11 months
Clinical Evaluation of Language Fundamentals (CELF-4) Pearson	In-clinic administration, direct assessment	Expressive and receptive language ability	Carriers participating in fMRI/MEG, and all controls, ages 5 to 21 years Non-familial adult and child controls *Note:* *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.*
CASL (Comprehensive Assessment of Spoken Language) Pearson	In-clinic administration, direct assessment	Spoken language	Carriers Non-carrier siblings Aged 3-21
CCC-2 (Children's Communication Checklist–2) Pearson	Paper questionnaire, parent/guardian report	Language and Pragmatic Skills	Carriers Non-carrier siblings Aged 4 years to 16 years 11 months who speak in full sentences

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Comprehensive Test of Phonological Processing — Non-Word Repetition task (CTOPP) Pro Ed Inc.	In-clinic administration, direct assessment	Phonological processing	Carriers Non-carrier siblings Non-carrier parents Aged 5 to 24 years
MacArthur Communicative Development Inventory — Words and Gestures, and Words and Phrases versions (MCDI) Pearson	Paper questionnaire, parent/guardian report	Early language use	Carriers Non-carrier siblings Aged 8 to 30 months or pre-phrase speech
Wechsler Individual Achievement Test, Version III (WIAT III) Pearson *Note: Only sections on reading comprehension, word reading, numerical operations, sentence composition (which includes sentence building and sentence completion) were completed.*	In-clinic administration, direct assessment	Academic achievement	All carriers in grade 1 up through age 50 years 11 months
Behavioral Inventories			
Adult Behavior Checklist (ABCL) ASEBA	Paper questionnaire, parent/spouse report	Problem behavior	Adult carriers, Adult non-carriers Aged 19 to 59 years
Child Behavior Checklist for ages 6 to 18 years (CBCL)	Paper questionnaire, parent/guardian report	Problem behavior	Carriers Non-carrier siblings Aged 6-18 years old

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Diagnostic Interview Schedule for Children (DISC) — Young Child and Child versions Columbia University, Casia Software	In-clinic administration or phone interview, direct assessment	Psychiatric disorders	Carriers Non-carrier siblings Aged 3-5 years 11 months (young child version) Aged 6-17 years 11 months (regular child version)
Symptom Checklist-90-Revised (SCL-90-R) Pearson	Paper questionnaire, self report	Psychiatric disorders	Adult carriers Adult Non-carriers
Broader Autism Phenotype Questionnaire (BAPQ) Piven	Paper questionnaire, self report	Broader autism phenotype	Adult carriers Adult Non-carriers
Behavior and Stereotyped Interest Questionnaire (BSIQ) Harvard University	In-clinic administration, direct assessment by a clinician who is reliable on the ADI (or trained by the author to administer this measure, if a site does not administer ADI)	Repetitive Behavior	Child carriers Child non-carrier siblings
Parenting Stress Index (PSI) WPS	Paper questionnaire, parent/guardian report	Parental stress	Parents (regarding all children <18 years)
Social Communication Questionnaire—Lifetime (SCQ) WPS	Paper questionnaire, parent/guardian report	Screen of ASD markers	Child carriers Child non-carriers Aged 4 – 17 years 11 months, with a mental age over 2 years

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Social Responsiveness Scale (SRS) WPS	Paper questionnaire, parent/guardian report	Autistic traits	Child carriers Child non-carriers Aged 4 – 18 years 11 months
Social Responsiveness Scale-Adult Research Version (SRS-ARV) WPS	Paper questionnaire, spouse/trusted adult report	Autistic traits	Carrier parents Non-carrier parents Carrier Adult children Non-carrier adult siblings Aged 19 years and up
Other Measures			
Dysmorphology—3dM D Pictures	Record form, 3-dimensional (3D) rendered photograph file	Broader autism phenotype	Carrier Adults Non-carrier Adults Carrier children Non-carrier children Only completed at Missouri and Harvard sites
Education History Interview Simons VIP	In-clinic administration, parent/self-report	Education history	Carrier adult - parents Carrier children
Medical History Interview (MHI — Adult and Child versions)	Telephone interview, parent/self-report	Family medical history	Carrier Adults Carrier children
Medication Interview	Telephone interview, parent/self-report	Current and past medication use	Carrier Adults Carrier children

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Mother Other Pregnancy Questionnaire	Paper questionnaire, self-report	Pregnancy history — non-live births	Adult carriers Adult-noncarriers Only completed by full-biological mothers
Previous Diagnosis Interview (Parent report on child, Self-Report Versions)	In-clinic administration	Previous health & mental health diagnoses	Adult carriers Child carriers
Puberty Questionnaire (versions for boys, girls, men, women)	Paper questionnaire, parent-report	Puberty status	Adult carrier Child carrier
Background History Form	Paper questionnaire, parent-report	Family demographic information, parent report of carrier's developmental milestones	Adult carrier Child carrier Adult non-familial controls Child non-familial controls
Feeding Questionnaire	Paper questionnaire, parent-report	Eating behavior	Carriers, siblings and controls, ages 13 years to 17 years 11 months
Eating in Absence of Hunger — Adult Self-Report, Parent Report on Child, and Child Self-Report versions (EAH)	Paper questionnaire, parent/self-report	Eating behavior	Adult carrier Adult non-carriers Child carriers Child non-carriers Adult controls Child controls Parents answer about themselves, children answer about themselves as able
LOC (Loss of Control) — Parent report on child, Self-report versions	Paper questionnaire. Self if possible, or parent about subject. Depending on cognitive age of child, the child may self-report. If a child self-report is obtained, a	Eating behavior	Adult carrier Adult non-carriers Child carriers Child non-carriers Adult controls Child controls Parents, siblings, controls and probands with cognitive age 60m+

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
	parent report on that child should also be obtained.		
Pediatric Sleep Questionnaire based on a sleep questionnaire by Ron Chervin, M.D.	Paper questionnaire, parent report	Sleep habits	Child carriers Child non-carriers (siblings) Child non-familial controls
Adult Sleep Questionnaire	Paper questionnaire, parent/self-report	Sleep habits	Parents adult carriers adult non-carrier siblings adult non-familial controls
Edinburgh Handedness Inventory Psychology Press	In-clinic administration, Direct assessment	Handedness	Adult carrier Adult non-carriers Child carriers Child non-carriers Adult controls Child controls ages > 2 years, use self-report 6 and up when able.
Height, Weight, Head Circumference Form (HWHC)	In-clinic administration; direct assessment	Physical characteristics	Adult carrier Adult non-carriers Child carriers Child non-carriers Adult non-familial controls Child non-familial controls
Neurological Exam	In-clinic administration; direct assessment	Cranial nerve and motor functioning, physician screen of speech production and quality, spine and skin exam	Child carriers Child non-carriers Child non-familial controls Adult Carriers Adult non-carriers
Neurologic Record Review Form (NRRF)	In-clinic administration; direct assessment	Medical history of neurological conditions	Child Carriers Adult Carriers

Instrument Name and Publisher	Measure Format	Measure Description	Administrations available by role
Structural MRI Scan Results Form	Record form completed by central imaging staff	Primary results from review of structural MRI scans (both pre-existing, if acceptable, and scan as part of the study)	Adult carriers Adult sibling non-carriers Child carriers Child non-carriers *Only those with structural MRIs

Simons Searchlight Genomic Data

There are two genomic datasets associated with Simons Searchlight. Simons Searchlight WGS1 (formerly Simons VIP WGS1). Simons Searchlight WGS1 includes WGS data for 21 individuals (6 families; 3 trios and 3 quads) where probands have 16p11.2 CNVs, was previously released on SFARI Base (accession ID: SFARI_SVIP_WGS_1) and corresponds to the publication Nuttle X. et al. Nature 536, 205-209 (2016).

Simons Searchlight WGS2 includes genetic data for 428 individuals, unique from WGS1 — 393 individuals with whole genome sequencing (WGS) and genome-wide SNP genotyping array data, as well as 35 individuals with SNP genotyping array data only. WGS data files include CRAMs, individual genomic VCFs (gVCFs), and project VCFs (pVCFs). SNP genotyping array data includes raw IDAT files as well as PLINK-formatted genotype calls. All data are mapped to human reference genome GRCh38, unless otherwise noted.

DNA was obtained from multiple sample source types, including whole blood, cell lines, and saliva (see column “dna_source” in the sample metadata table). All samples were processed using the same sequencing methods and with two Illumina genotyping array versions, described in more detail in the “Sequencing Methods” and “Genotyping Methods” sections of the Simons Searchlight WGS Release Notes.

Research Match

Research Match is a service that enables approved researchers to recontact participants from SFARI autism cohorts, including Simons Searchlight, for recruitment into new research studies. Researchers may be approved to recruit participants for in-clinic or remote studies, or to utilize the Research Match platform to build their online surveys. To date, over 3,000 families participating in Simons Searchlight have been invited to join a study through Research Match, and approximately 1,750 families have responded to Research Match invitations.

Please note that applications to access the Research Match service must be submitted on SFARI Base. Click here for more information on using Research Match services.

How to access SFARI Base and request SFARI resources

1. Log into SFARI Base here. If you do not have a SFARI Base account, you may create one. As part of account creation, you will need to affiliate with an institution. If your institution is not registered on SFARI Base, you will be prompted to provide institution details as part of registration. This includes the name and email address for a Signing Official who has authority to represent your institution in a legal agreement, similar to a material transfer agreement.

2. Review your institution's executed Researcher Distribution Agreement (RDA) and agree to the joinder to the RDA. 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 agree to the joinder agreement to the RDA for their respective institution.

   For institutions that do not yet have an executed RDA, it will be sent to the institutional officer for review and signature upon approval of a request in SFARI Base.

3. Create a SFARI Base project, which includes the title, abstract, and an IRB approval or exemption document. 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.

4. Create a SFARI Base request, which includes specifying the type of resource you would like to request access to (phenotype, genetic, or imaging data; biospecimens; and/or research match application). The submitted request will be reviewed by the SFARI science team or appropriate committee.

Simons Searchlight Sample Consent Forms

Phase 1(Simons VIP)

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

Phase 2

https://simonsfoundation.s3.amazonaws.com/share/Simons_Searchlight/simons_searchlight_consent_form.pdf

Simons Searchlight Acknowledgment Policy

All SFARI-funded investigators who use Simons Searchlight phenotypic data, genetic data, imaging data and/or biospecimens:

- "This work was supported by a grant from SFARI (Grant Number, Awardee Initials)."
- We are grateful to all of the families at the participating Simons Searchlight sites as well as the Simons Searchlight Consortium, formerly the Simons VIP Consortium.
- We appreciate obtaining access to **[include here the type of data used, eg. phenotypic, genetic and/or imaging]** data on SFARI Base.
- Approved researchers can obtain the Simons Searchlight population dataset described in this study **([include here the URL of the population used, obtained from SFARI Base])** by applying at https://base.sfari.org.

All non-SFARI-funded investigators who use Simons Searchlight phenotypic data, genetic data, imaging data and/or biospecimens:

- We are grateful to all of the families at the participating Simons Searchlight sites as well as the Simons Searchlight Consortium, formerly the Simons VIP Consortium.
- We appreciate obtaining access to **[include here the type of data used, eg. phenotypic, genetic and/or imaging]** data on SFARI Base.
- Approved researchers can obtain the Simons Searchlight population dataset described in this study **([include here the URL of the population used, obtained from SFARI Base])** by applying at https://base.sfari.org.

If you have additional questions, email sdbr@simonsfoundation.org.