Demographic and Clinical Characteristics of Participants in the Australian Autism Biobank

Gail A. Alvares,[1,2] on behalf of the Australian Autism Biobank team

1. Cooperative Research Centre for Living with Autism (Autism CRC), Long Pocket, Brisbane, Australia; 2. Telethon Kids Institute, University of Western Australia, Perth, Australia

Background

- There is enormous clinical (phenotypic) and genetic heterogeneity within individuals diagnosed with Autism Spectrum Disorder (ASD);
- Alongside significant advances in genetic and biological research have been increasing imperatives to establish large bio-resources to support discovery research;
- To date, there has not been a single collective effort to establish such a resource in Australia, which has unique ethnic and cultural diversity.
- The Australian Autism Biobank was initiated in 2014 by the Cooperative Research Centre for Living with Autism (Autism CRC).

Objectives

- To describe the demographic and clinical characteristics of participants in the Australian Autism Biobank

	ASD n = 1151	ASD-Q n = 17	Sibling n = 263	Control n = 150
Female(%)	251 (25.81%)	9 (52.94%)	136 (51.71%)	76 (50.67%)
Age	7.48 (3.87)	6.22 (2.56)	8.12 (4.21)	6.25 (3.37)
Age at dx.	4.54 (2.81)	-	-	-
Mat. Ethnicity (%)				
Caucasian	766 (81.4%)	7 (100%)	90 (84.1%)	97 (91.5%)
Asian	111 (11.8%)	0 (0%)	8 (7.5%)	8 (7.5%)
Aboriginal/Maori	20 (2.1%)	0 (0%)	4 (3.7%)	1 (0.9%)
Other	44 (4.7%)	0 (0%)	5 (4.7%)	0 (0%)
Pat. Ethnicity (%)				
Caucasian	778 (83.4%)	7 (100%)	91 (84.3%)	87 (85.3%)
Asian	100 (10.7%)	0 (0%)	12 (11.1%)	116 (10.1%)
Aboriginal/Maori	20 (2.1%)	0 (0%)	3 (2.8%)	28 (2.4%)
Other	35 (3.8%)	0 (0%)	2 (1.9%)	43 (3.7%)

Methods

See study protocol : *Alvares et al., 2018 BMC Pediatrics*

Participants

(1) children diagnosed with ASD, and both biological parents (where possible);
(2) children referred for an ASD diagnosis, but not meeting criteria ('ASD-Query');
(3) siblings of children diagnosed with ASD;
(4) children without an ASD diagnosis ('controls').
No exclusion criteria regarding language level, cognitive ability, or medical, psychiatric or genetic conditions (other than ASD).

Phenotypic Data

Clinical assessments:
- Autism Diagnostic Observation Schedule
- Mullen Scales of Early Learning or Wechsler Intelligence Scale for Children
Questionnaires:
- Family and medical history
- Vineland Adaptive Behavior Scales – 2nd ed.

Age at Participation

Results

Sample Characteristics

- Sex distribution was 3.51:1 (male:female) in the ASD group, but equivalent in other groups.
- The cohort included:
  - 588 trios (samples from child diagnosed with ASD, plus mother and father) and 134 quads (plus a non-diagnosed sibling);
  - 334 children from multiplex families (between 2-5 children diagnosed);
  - 20 concordant (both children diagnosed) and 13 discordant (one child diagnosed) twin pairs
- Ethnic distribution
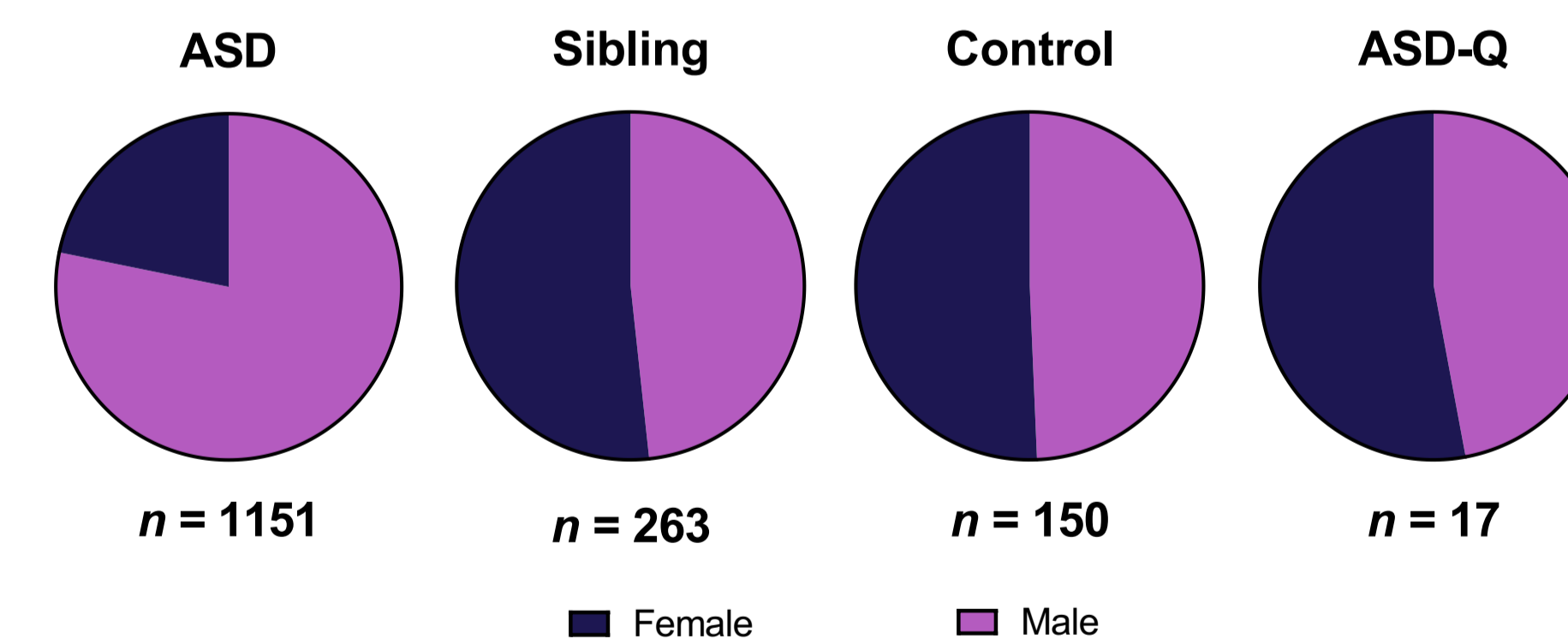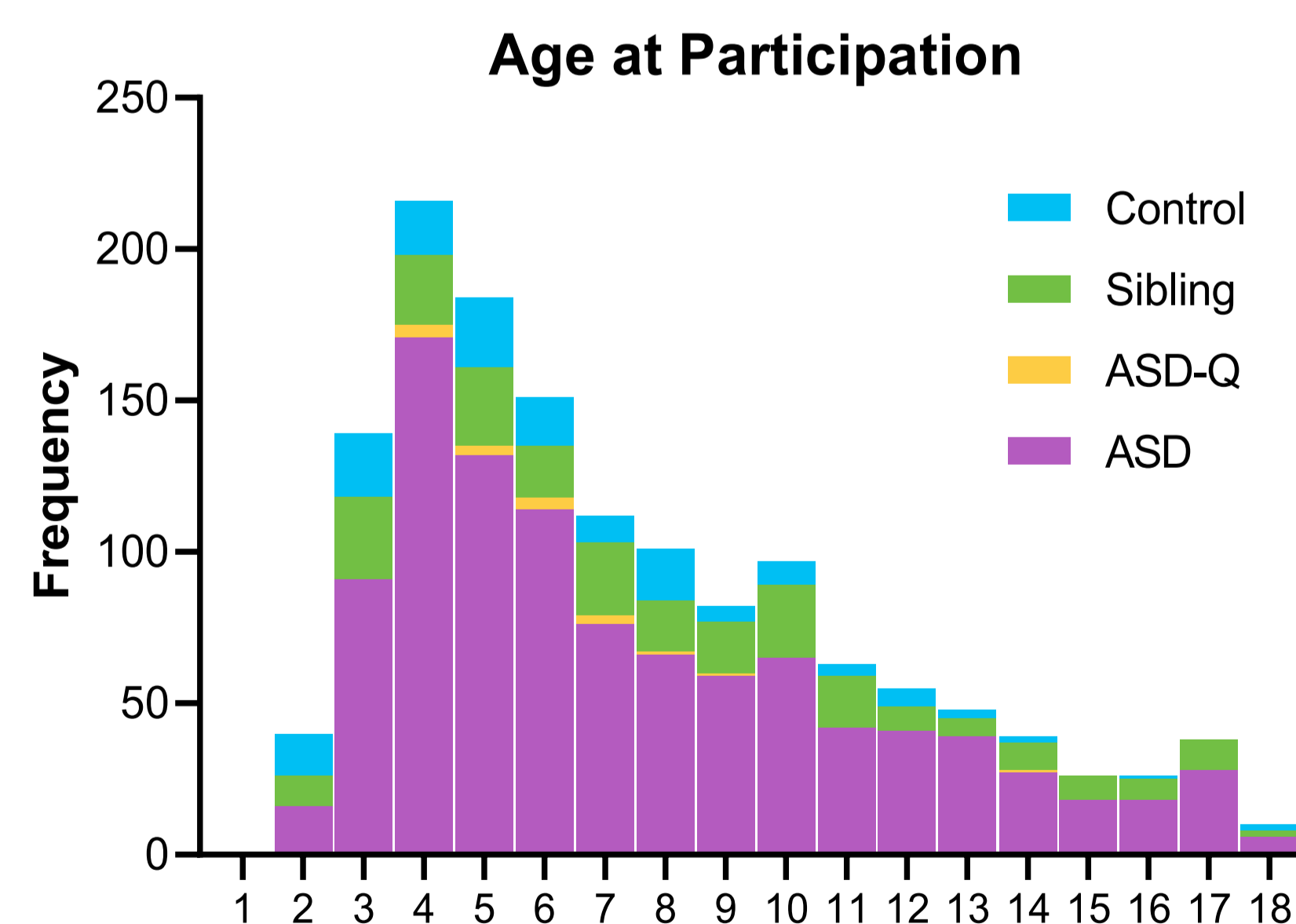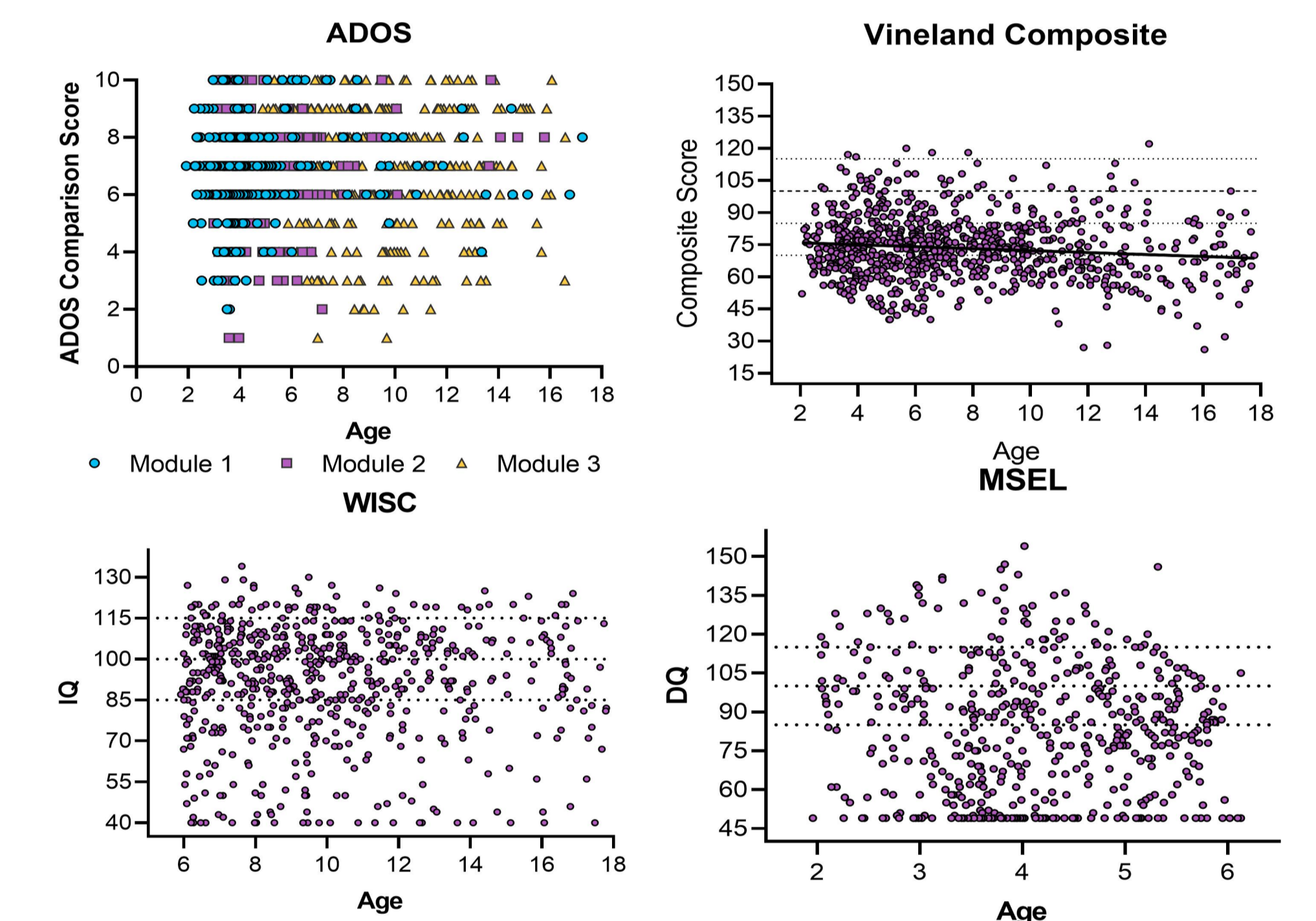 - 83% Caucasian, 11% Asian, 2% Aboriginal, Maori or Torres Strait Islander, 4% Other

Within the ASD group:

- Age at ASD diagnosis: 1-17 years (4.54±2.81)
- 27.18% reported a diagnosed Intellectual Disability or Developmental Delay (n=274)
- 4.76% reported epilepsy (n=48), 5.16% ADHD (n=52), and 2.18% a genetic condition (n=22).

ASD	Sibling	Control	ASD-Q
n = 1151	n = 263	n = 150	n = 17

■ Female ■ Male

Results

ASD Clinical Characteristics

- IQ scores ranged from 40-134 (84.81±22.49); DQ scores ranged from 49-138 (73.11±22.53)
- Vineland scores ranged from 26-122 (73.34±14.67), associated with age (r=-.13**)

Discussion

- This resource is currently the largest biological and clinical repository about ASD in Australia.
- The combined database contains detailed demographic and phenotypic (clinical) data matched to biological samples.
- Data access requests are now open for applications: autismcrc.com.au/biobank

For more information

Dr Gail Alvares | Email: Gail.Alvares@telethonkids.org.au

autismcrc.com.au
@autismcrc