



# Factors that Promote Early ASD Diagnosis among Simplex Families in an Urban Setting

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## Introduction

- Age at ASD Diagnosis**
  - ASD can be reliably diagnosed in children as young as 2 years (Steiner, Goldsmith, Snow, and Chawarska, 2012) and some ASD symptoms can be detected as early as 6 months of age (Chawarska and Shic, 2013)
  - Age at diagnosis still varies greatly, with a median age of 4.5 years (CDC, 2014)
- Early Diagnosis**
  - Early diagnosis and intervention significantly and positively impact child outcomes (Zwaigenbaum et al., 2015)
  - Little is known about biological and behavioral factors that may impact early diagnosis (Herlihy et al., 2015; Liptak et al., 2008)
- Autism Genetics**
  - Recent advances have been made in the understanding of genetic factors implicated in autism
  - Growth in knowledge of autism genetics necessitates an examination of relationship between individuals' genetic status and age at autism diagnosis
  - Likely Gene-Disrupting Mutation (LGD): genetic mutation that results in a shortened gene product (Iossifov et al., 2015)
    - 42% of de novo LGDs estimated to account for 9% of ASD diagnoses (Iossifov et al., 2014)

## Objective

To determine the factors that promote early ASD diagnosis for children in simplex families within a homogeneous socio-demographic community.

## Methods

**Sample:** Simons Simplex Collection (SSC) (University of Washington site only) (Fischbach and Lord, 2010)

- SSC: Repository containing phenotypic and genotypic data and biospecimens from 2,700 simplex families
- N = 182 children (79.9% male, mean age = 9.41 years)
- 28 children with an identified Likely Gene-Disrupting Mutation (LGD), 154 without (Iossifov et al., 2015)

**Variables**

Child variables: presence of an LGD, developmental regression, and intellectual ability, age at parental concern

Demographic variables: gender, maternal education, family income, race

**Measures**

Age at diagnosis: SSC participant screening interview

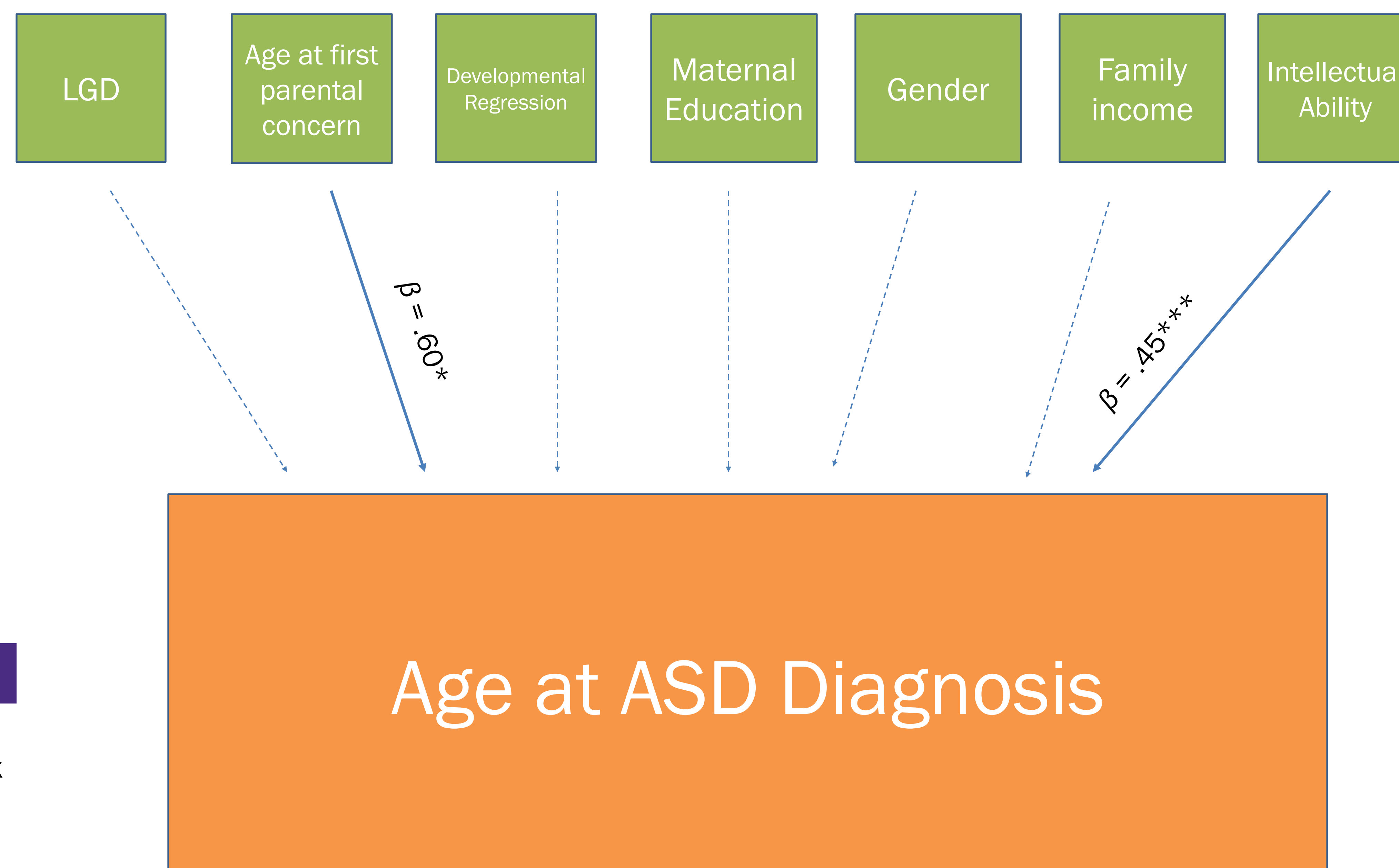
Developmental regression, age at parental concern: Autism Diagnostic Interview, Revised (Lord et al., 1994)

Intellectual ability: Differential Ability Scales (DAS) or Mullen Scales of Early Learning (Elliott, 1990; Mullen, 1995)

**Analysis**

Multiple linear regression was performed to determine whether there were main effects of variables of interest upon age at ASD diagnosis.

## Results



## Results Continued

### Promotive Factors for Early Identification and Diagnosis of ASD:

- Main effect of intellectual ability on age at diagnosis over and above all other variables of interest
  - Strong positive relationship between IQ and age at diagnosis
- Significant effect of age at first parental concern on age at diagnosis
  - Positive relationship between parental concern and age at diagnosis
- Minor trend-level effect of presence of developmental regression on age at diagnosis
- Demographic and child variables that did not have a significant impact upon age at diagnosis were child's gender, race, maternal education, and family income
- Presence or absence of an LGD mutation also did not impact age at diagnosis

## Discussion

**Objective:** To determine the factors that promote early ASD diagnosis for children in simplex families within a homogeneous socio-demographic community.

- This analysis selected for an urban sample within a relatively high-income area with relatively equal access to diagnostic services.
- In a homogenous socio-demographic community, children's intellectual ability may impact when children are diagnosed.
- Though parental concern has not been shown to promote early ASD diagnosis in previous literature, age at first parental concern does appear to contribute to age at diagnosis in this sample.
- Importance of using a simplex sample:
  - Having an older sibling with ASD has been shown to promote early diagnosis, so focusing on families with only one child with ASD allows us to examine the factors that impact diagnosis when families do not have previous experience with ASD.

### Implications and Future Directions:

- If intellectual ability, a characteristic not specific to ASD, impacts age at diagnosis when environmental factors are homogeneous, parent education is needed to help parents and caregivers recognize symptoms specific to ASD. This is especially important for children with autism who do not exhibit intellectual impairments.
- A larger group of individuals with an identified LGD is needed to more closely examine the impact of LGD status on age at diagnosis.
- Further examination of the impact of maternal education, family income, and other family variables upon age at first parental concern is also necessary- do demographic characteristics have an impact upon family awareness of ASD symptomatology in this sample?

<sup>†</sup> signifies <.10; \* signifies <.05; \*\* signifies .01; \*\*\* signifies <.001