



# Behavioral Health of People with Mosaic Down Syndrome

Ruth C. Brown, PhD<sup>1</sup>, Rebekah Zee, MS<sup>2</sup>, Quinn Hurshman, MS<sup>2</sup>,

Ananda B. Amstadter, Ph.D.<sup>1</sup>, Colleen Jackson-Cook, Ph.D.<sup>3</sup>

<sup>1</sup> Department of Psychiatry, Virginia Commonwealth University

<sup>2</sup> Department of Human and Molecular Genetics, Virginia Commonwealth University



## Introduction

Mosaic Down syndrome (mDS) is a variant form of Down syndrome (DS) in which an individual has at least two chromosomally distinct cell lines that are derived from a single zygote and occurs in approximately 1.3-5% of people diagnosed with DS<sup>1</sup>. There is evidence to suggest that the percentage of trisomic cells in persons with mDS is associated with the severity of phenotypic traits (e.g., IQ, epicanthal folds, congenital heart defects)<sup>2</sup>. The association between percentage of trisomic cells and behavioral phenotypes has not been explored.

## Objectives

To examine the prevalence of self-report and caregiver-report measures of depression, anxiety, and stress among participants with mDS and to examine the association of these behavioral health outcomes with percent trisomy from an ongoing study.

## Methods

Data was drawn from the Life Experience and Feelings (LEAF) Study. Enrollment took place at several DS-focused conferences and travel to large US cities. A total of 30 participants have participated to date.

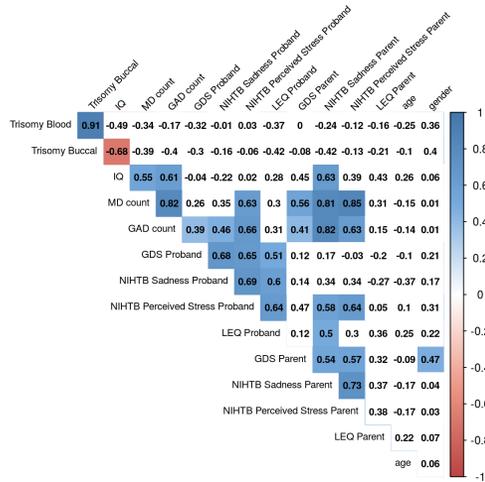
## Measures

- **IQ** was estimated using the Kaufman Brief Intelligence Test (KBIT-2)
- **Major Depression (MD)** and **Generalized Anxiety Disorder (GAD)** DSM-5 symptom count assessed using the Psychiatric Assessment Schedule for people with IDD (PAS-ADD)<sup>3</sup>.
- **Stressful life events** were identified using self-reported and parent-reported Life Events Questionnaire (LEQ)<sup>4</sup> # of past year events rated as having a "bad" effect.
- **Glasgow Depression Scale (GDS)**<sup>5</sup>, a self-report ( $\alpha = .73$ ) and parent-report ( $\alpha = .73$ ). Clinical cutoff for self-report =13<sup>5</sup>.
- **The NIH Toolbox (NIHTB)**<sup>6</sup> **Sadness** scale self-report and parent proxy battery were used to provide a norm-referenced screen for depression, and the **Perceived Stress** scale provides a norm-referenced screen for stress. Clinical cutoff = T-score 70.

**Table 1. Descriptive Statistics**

Variable (available n)	Mean (SD)
Age (n = 30)	22.7 (10.3)
Gender, male (n = 30)	12 (40.0%)
IQ (n = 19)	75.9 (15.3)
Verbal IQ (n = 19)	74.4 (12.4)
Nonverbal IQ	82.4 (17.2)
GDS Proband (n = 26)	14.0 (7.29)
GDS Parent (n = 27)	7.85 (4.28)
Percent Trisomy Blood (n = 22)	27.5 (32.2)
Percent Trisomy Buccal (n = 20)	46.1 (22.1)
DSM-5 MD Symptoms (n = 30)	1.60 (2.03)
DSM-5 GAD Symptoms (n = 30)	1.53 (1.98)
NIHTB Sadness Proband (n = 21)	56.8 (13.6)
NIHTB Sadness Parent (n = 25)	57.2 (13.4)
NIHTB Perceived Stress Proband (n = 19)	56.9 (8.85)
NIHTB Perceived Stress Parent (n = 25)	59.2 (8.57)
LEQ # Stressful Events Proband (n = 24)	3.54 (2.95)
LEQ # Stressful Events Parent (n = 27)	2.22 (2.65)

**Figure 1. Correlations between percentage trisomy, IQ, and behavioral health symptoms. Shaded regions represent correlations significant at  $p < .05$ .**



## Results

See Table 1 for means and standard deviations of study variables. Figure 1 displays correlations between study variables.

Percent trisomy in buccal was significantly associated with IQ. IQ, but not percent trisomy, was statistically associated with MD and GAD diagnoses such that higher IQ was associated with poorer behavioral health.

On the Glasgow Depression Scale, 13 probands, 54%, reported clinically significant depression symptoms. Diagnosis of Depression and Anxiety with the PAS-ADD.

On the PAS-ADD, participants reported a  $M = 1.6$ ,  $SD = 2.03$  DSM-5 depression criteria, and 2 met DSM-5 criteria for depression. Participants endorsed  $M = 1.53$ ,  $SD = 1.98$  DSM-5 GAD criteria, and 1 met DSM-5 criteria for GAD.

Scores on the NIHTB Sadness proband report were significantly higher than the NIH Toolbox normative sample ( $t(20) = 2.27$ ,  $p = 0.03$ ). 19% ( $n = 4$ ) reported clinically significant sadness (i.e., T-score  $\geq 70$ ). Scores on the parent-rated NIHTB Sadness were also significantly higher than the NIH Toolbox normative sample ( $t(24) = 2.67$ ,  $p = 0.01$ ). 20% ( $n = 5$ ) reported clinically significant sadness.

Scores on the NIHTB Perceived Stress proband report were significantly higher than the NIH Toolbox normative sample ( $t(18) = 3.42$ ,  $p < 0.01$ ). 5% ( $n = 1$ ) reported clinically significant symptoms of stress (i.e., T-score  $\geq 70$ ). Scores on the parent-rated NIHTB Perceived Stress were significantly higher than the NIH Toolbox normative sample ( $t(24) = 5.34$ ,  $p < .01$ ). 20% ( $n = 5$ ) reported clinically significant stress.

## Conclusions

Evidence across measures (diagnostic and screening) suggests that people with mDS experience symptoms of depression and sadness at a greater rate than the general population. 6% met DSM-5 criteria for MD; the population prevalence has been estimated at 2.6% for past month MD<sup>7</sup>. 3% met DSM-5 criteria for GAD; the population prevalence has been estimated at 0.8% for past-month GAD<sup>8</sup>.

## Conclusions, Continued

Scores on the Glasgow Depression Scale (developed for people with intellectual disability) and the NIH Toolbox (developed for the general population) suggested high rates of positive screens for depression, sadness, and perceived stress.

Correlation analyses did not find a statistically significant effect of trisomy percentage on behavioral health outcomes, although correlation coefficients were of small-medium effect size and the direction of effect suggested that those with less trisomy exhibited higher levels of stress and poorer behavioral health.

The correspondence between parent and proband report was generally high (compared to average parent-child correlations in general population children of  $\sim .20$ ), but divergent enough to suggest unique information is conveyed by informant type. Together with the convergent validity of proband report across measures, these findings highlight the need to include self-report in mental and behavioral health assessments when possible<sup>10</sup>.

Although there was variability in the respondent and measurement type, stress exposure and symptoms were positively associated with behavioral health symptoms, consistent with findings from the general population.

## Limitations and Future Directions

Statistical analyses are underpowered due to small sample size. Data collection is ongoing, but delayed due to COVID-19 pandemic. Future analyses will examine the associations between stress exposure and mental health and test for moderation effects of trisomy. Additional work is planned to understand predictors of parent-proband discrepancies (e.g., age, IQ, supervision) to inform interpretation of parent and proband reports. This study will inform adaptation of behavioral health assessments for use in people with non-mosaic DS planned over the next two years. Future studies will compare the rates behavioral health among people with mDS and non-mosaic DS.

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