The background features a watercolor-style wash in various shades of blue and teal. On the right side, there is a faint, light-colored silhouette of a human brain, facing right. The overall aesthetic is clean and professional.

# EXECUTIVE AND ADAPTIVE FUNCTION IN ADULTS WITH MAPLE SYRUP URINE DISEASE

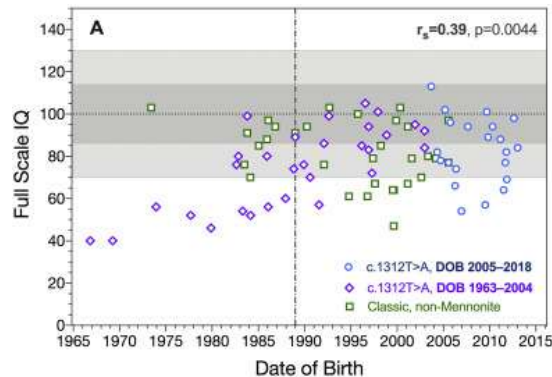
JESSICA GOLD, MD, PHD

JUNE 27, 2024

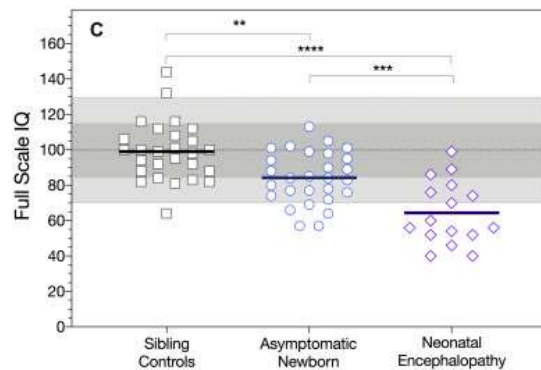
# OBJECTIVES

- To correlate medical self-management, independent living skills, and executive and adaptative function in adults with MSUD
- To characterize current social outcomes for adults with MSUD
- To identify gaps in preparation for health care transition

# NEUROCOGNITIVE OUTCOMES IN MSUD



- Strauss et al: Full-scale IQ correlates with birthdate and presence of neonatal encephalopathy
  - Mean age 18.5 years



- Bouchereau et al: Normal full scale IQ in but higher verbal than performance IQ suggestive of executive dysfunction
  - Mean age: 8.7 years



# EXECUTIVE FUNCTIONING EFFECTS DAILY LIFE

## Mental Health

- EF impaired in ADHD, depression, obsessive compulsive disorder, and generalized anxiety disorder

## Physical Health

- Poor EF associated with poor treatment adherence, disordered eating, and substance abuse

## Education

- EF is more important for school readiness than IQ

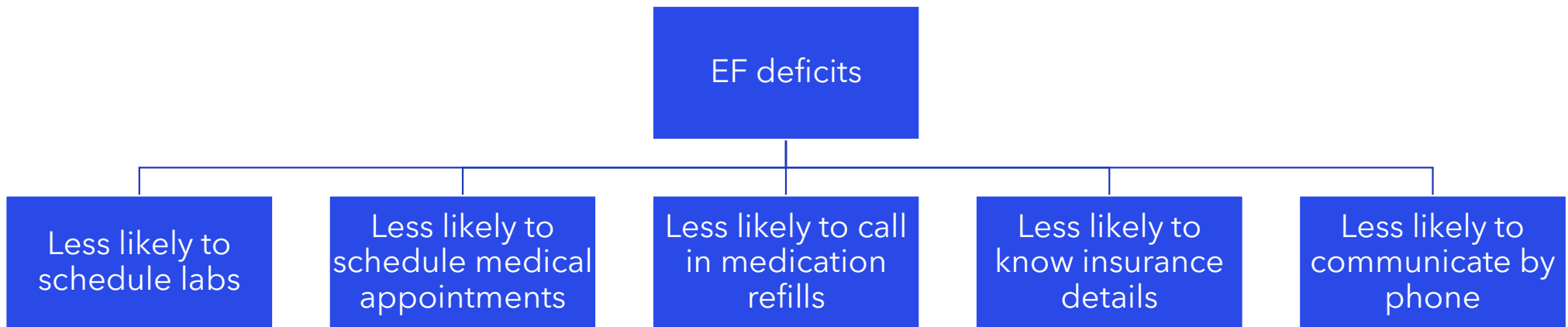
## Job Success

- EF associated with productivity. Poor EF correlates with difficulty finding and keeping a job

## Quality of Life

- Better EF = Better Quality of Life

# EF CORRELATES WITH MEDICAL SELF-MANAGEMENT SKILLS



Less likely to transfer care to adult-oriented provider

# STUDY DESIGN

Neurocognitive  
Assessment on Executive  
and Adaptive Function

Quality of Life and ADLs  
Measurements

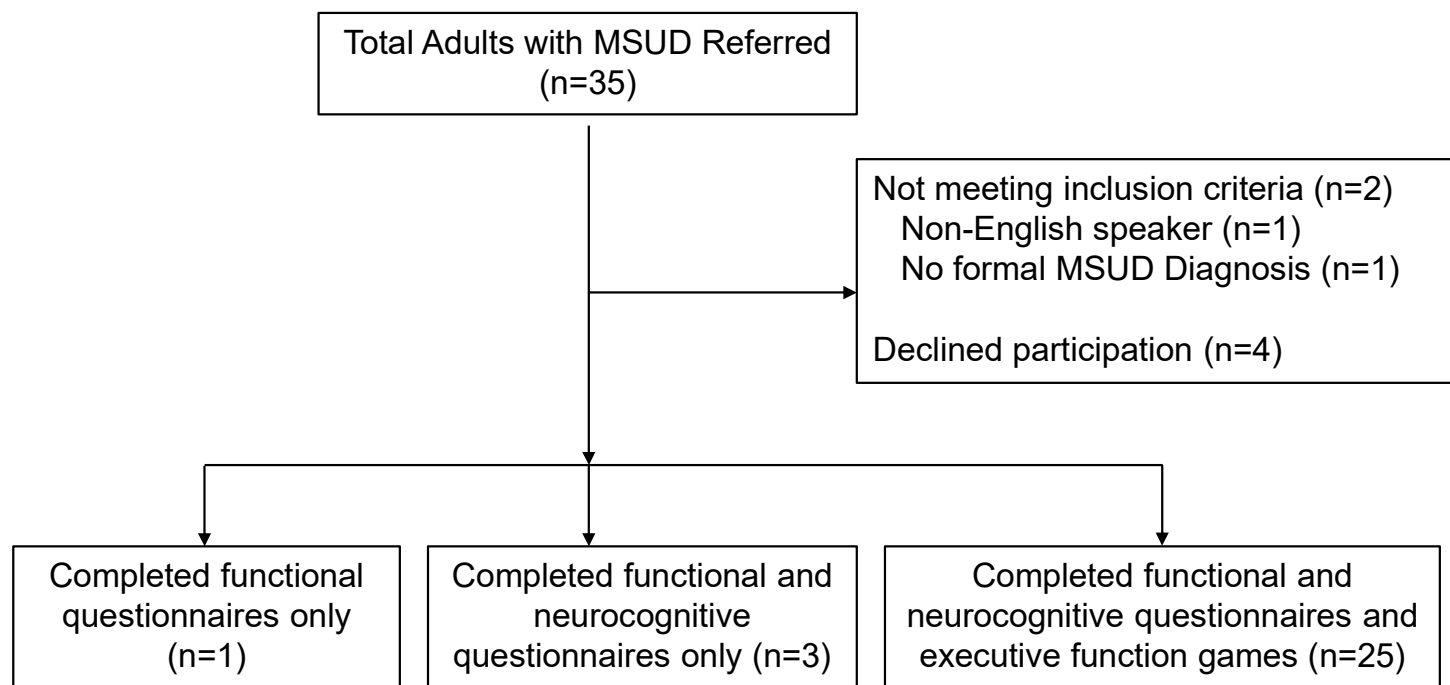
Semi-Structured Interviews

Improved instruments for Transition Readiness  
More focused support for adult life  
Guidance for prognostication

Enrollment Criteria:

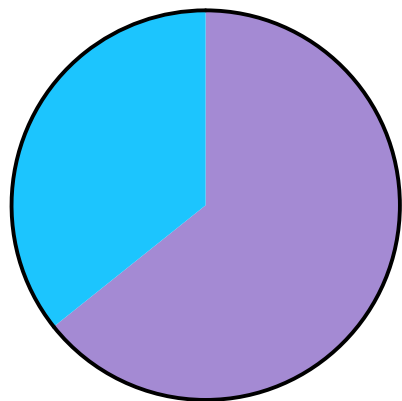
- Age 21+
- English-speaking
- Access to computer/tablet/smart phone

# STUDY ENROLLMENT



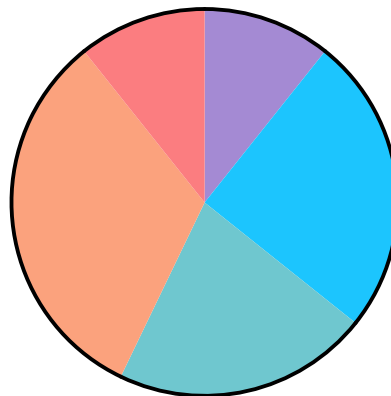


# DEMOGRAPHICS



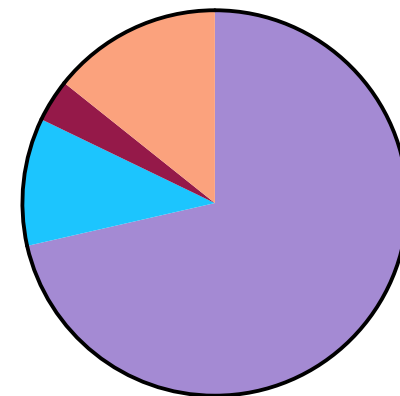
Total=28

Female  
Male



Total=28

21-25  
26-30  
31-35  
36-40  
41+



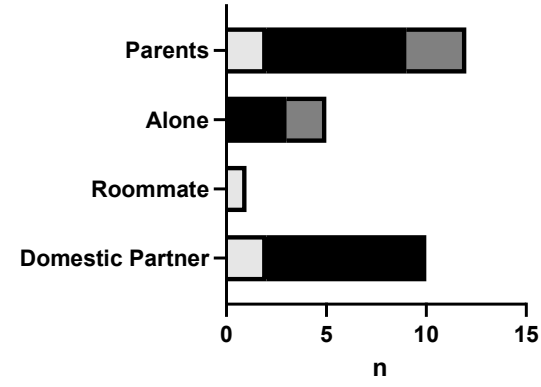
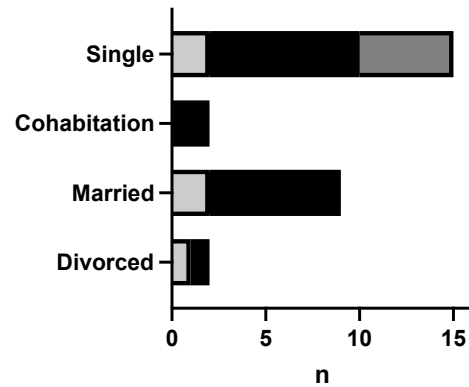
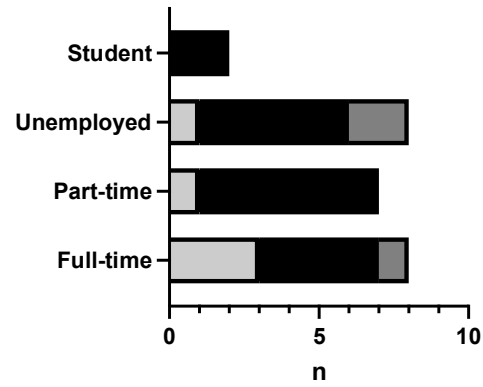
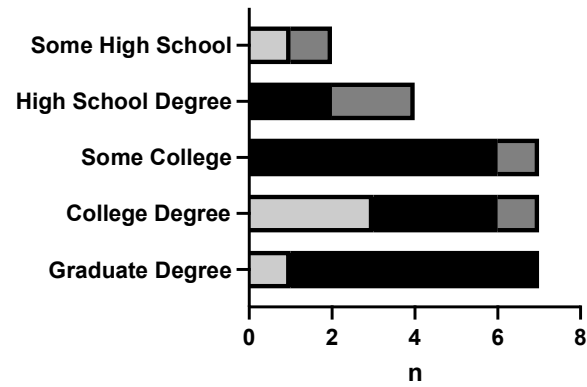
Total=28

White  
Black  
Asian Indian  
Latinx

24 participants (85.8%) were symptomatic at diagnosis  
5 participants (17.8%) had liver transplantation



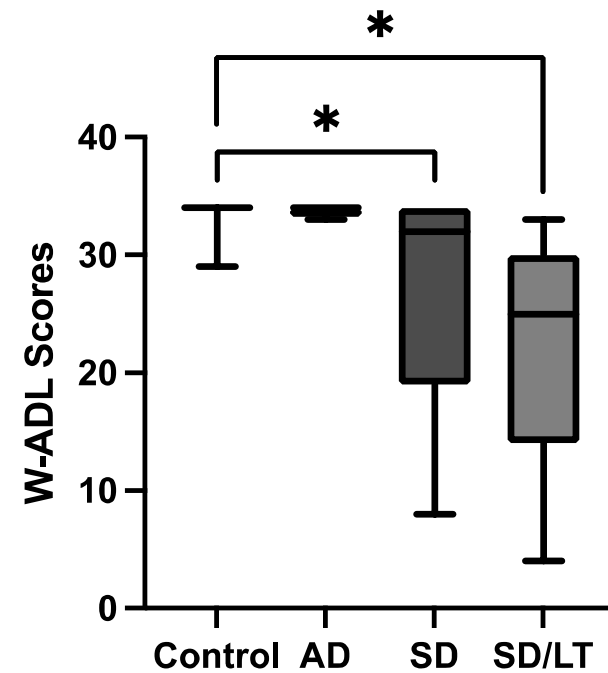
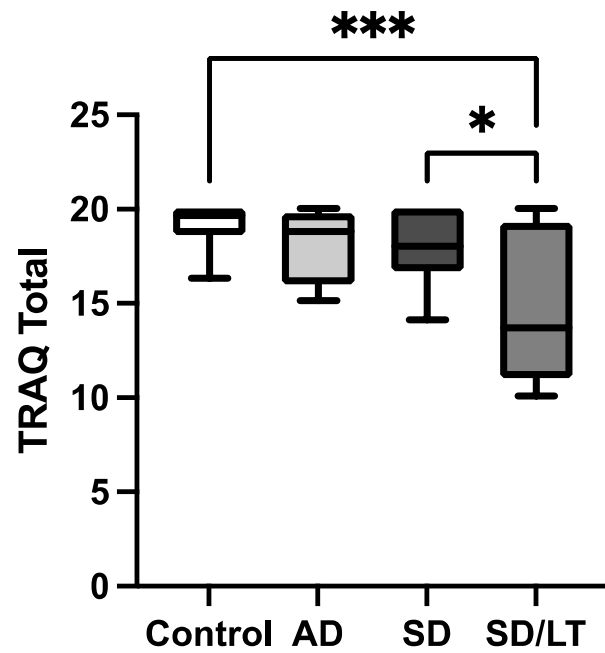
# LONG-TERM OUTCOMES



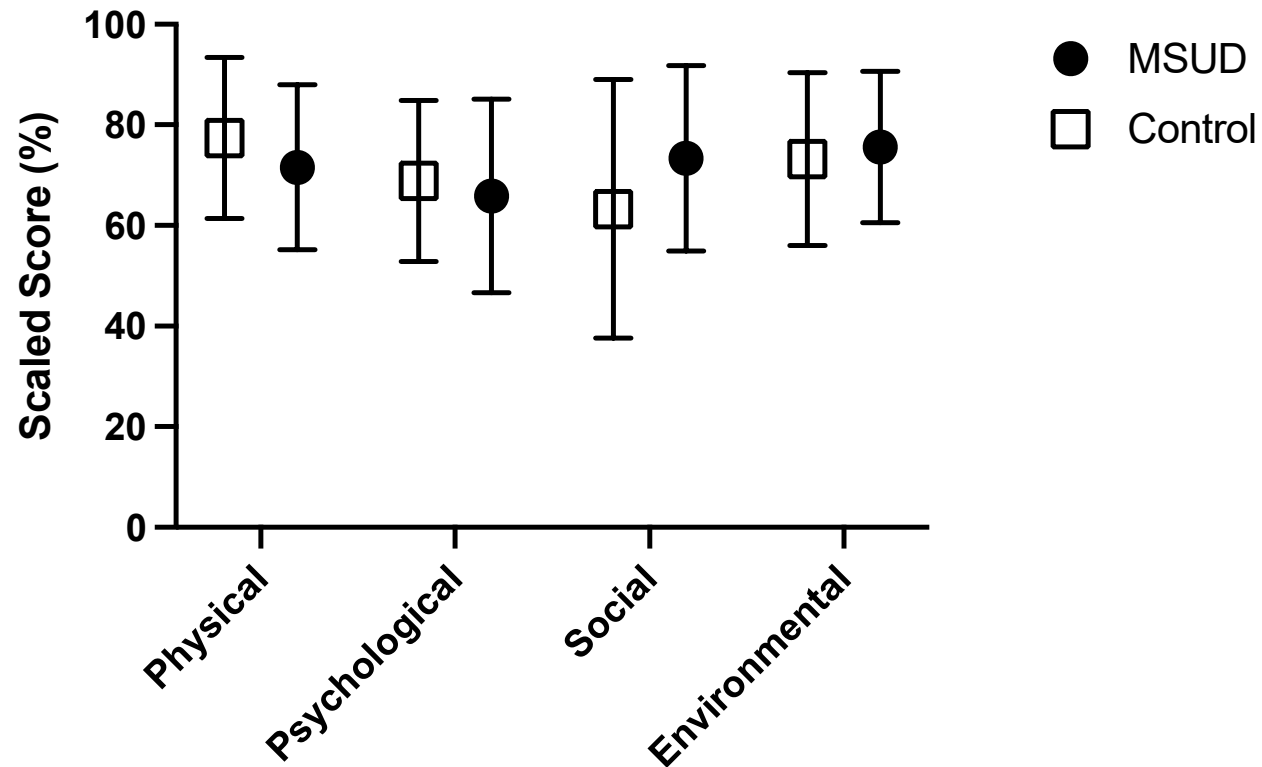
Asymptomatic Diagnosis
  Symptomatic Diagnosis
  Symptomatic Diagnosis, Liver Transplant



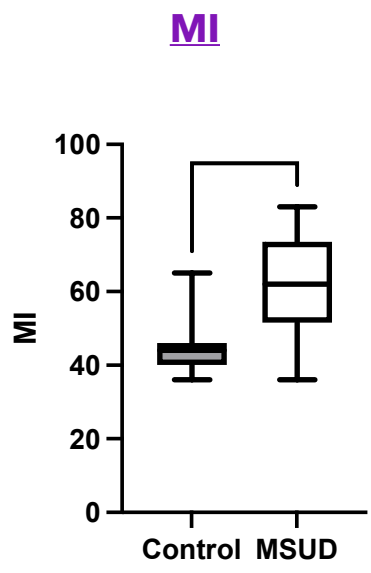
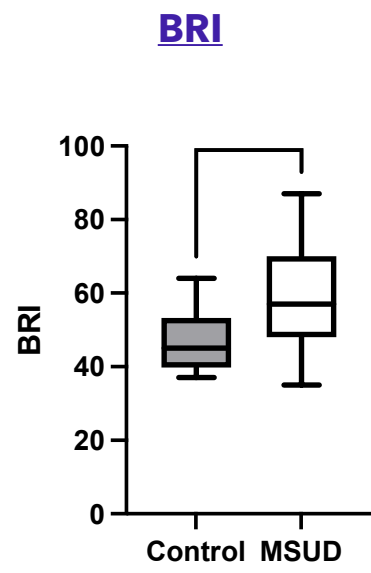
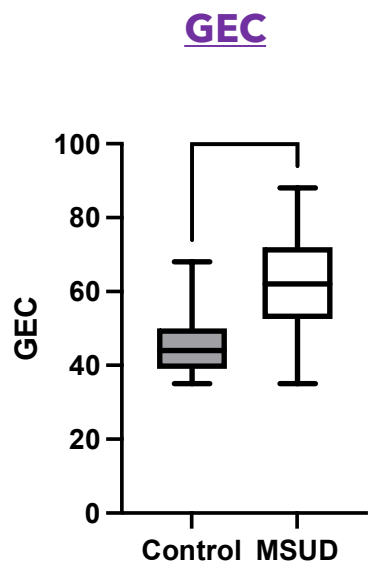
# SKILLS FOR INDEPENDENCE



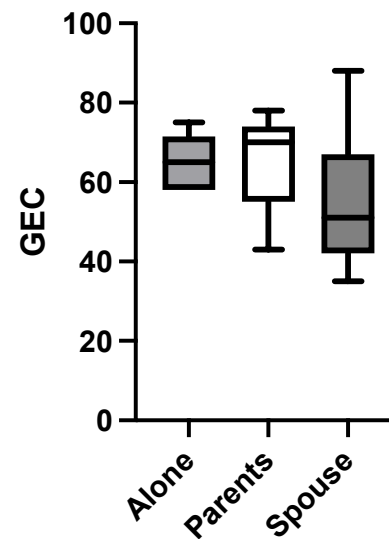
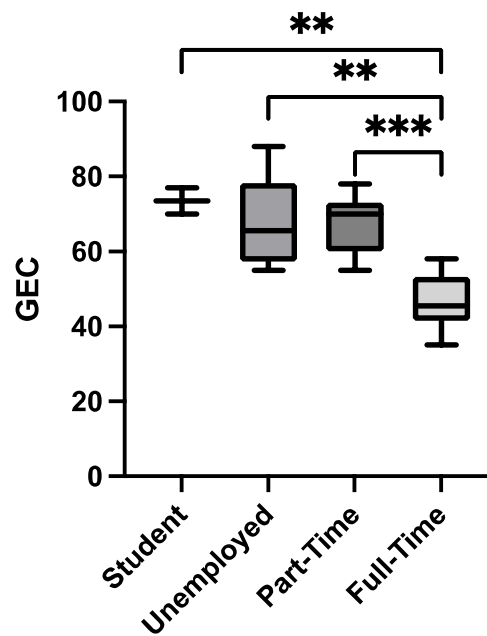
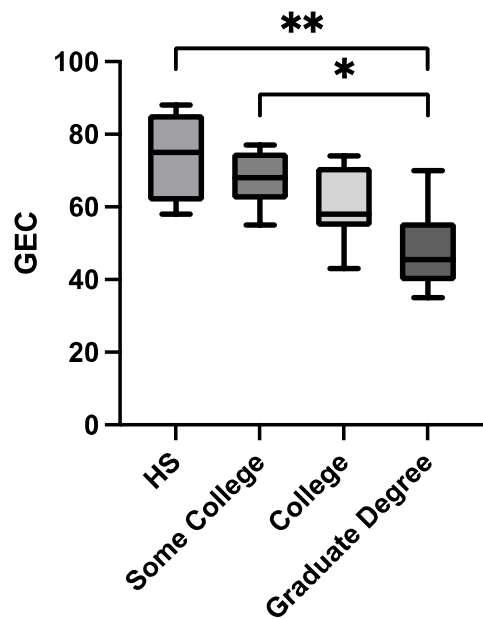
# SELF-REPORTED QUALITY OF LIFE IS GOOD



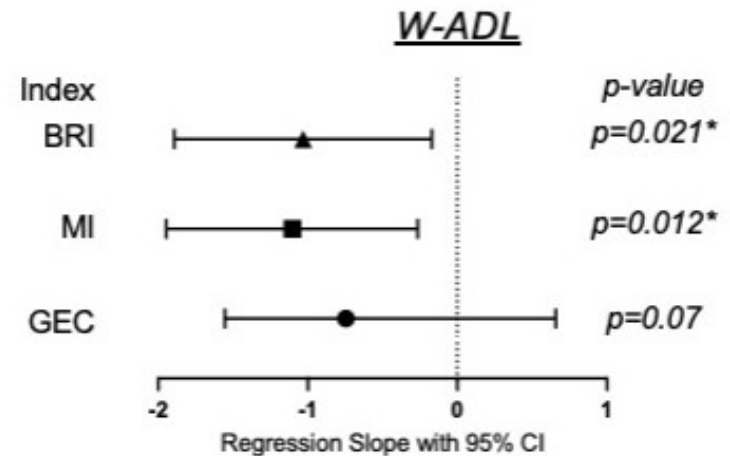
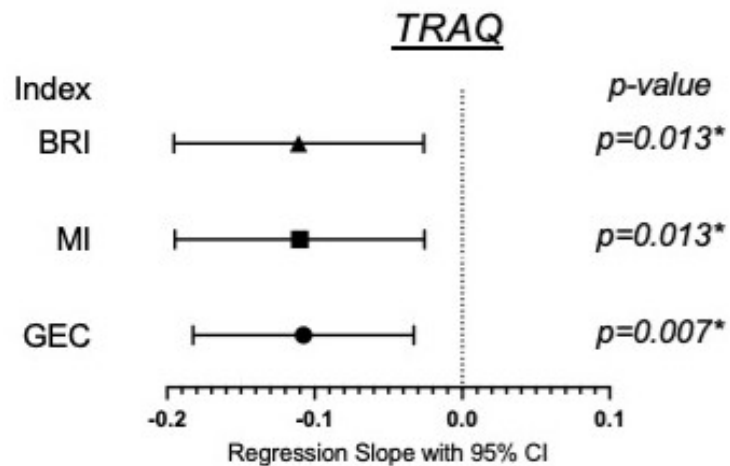
# BRIEF-A: EXECUTIVE FUNCTION QUESTIONNAIRE



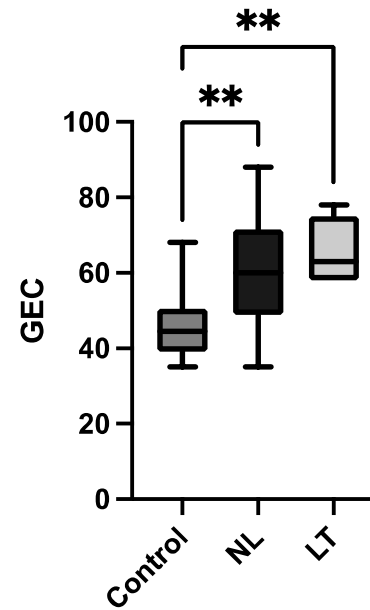
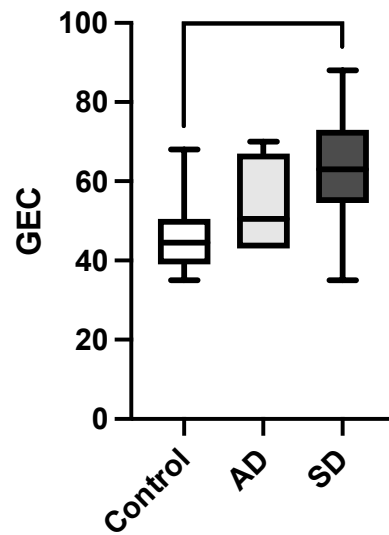
# BRIEF-A SCORES CORRELATED WITH EDUCATION AND EMPLOYMENT



# BRIEF-A SCORES CORRELATED WITH INDEPENDENT LIVING SKILLS

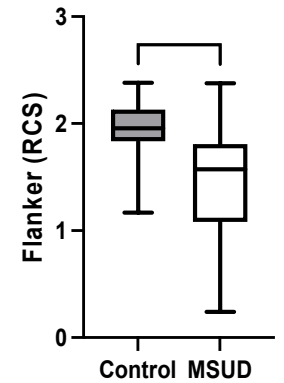
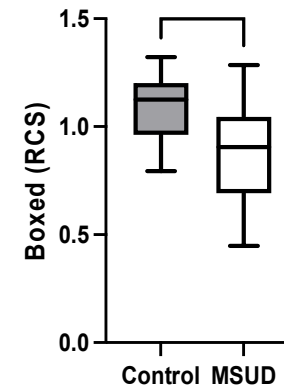
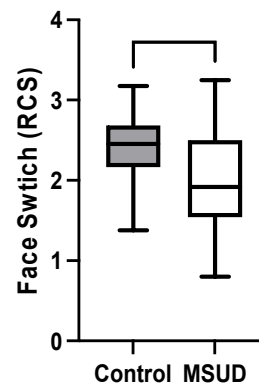


# CLINICAL HISTORY MAY IMPACT BRIEF-A SCORES

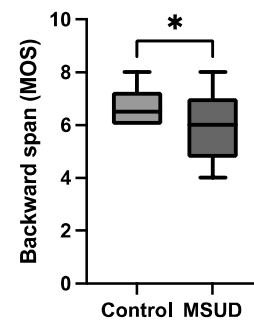
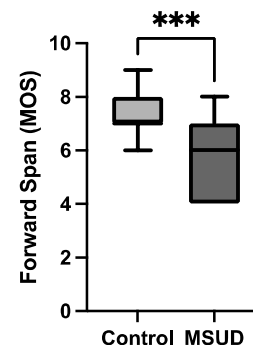
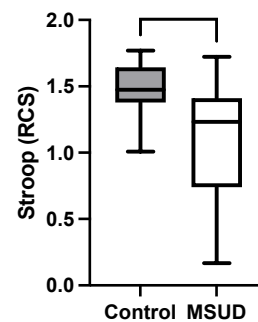
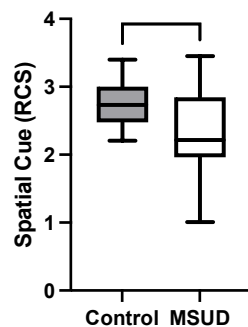
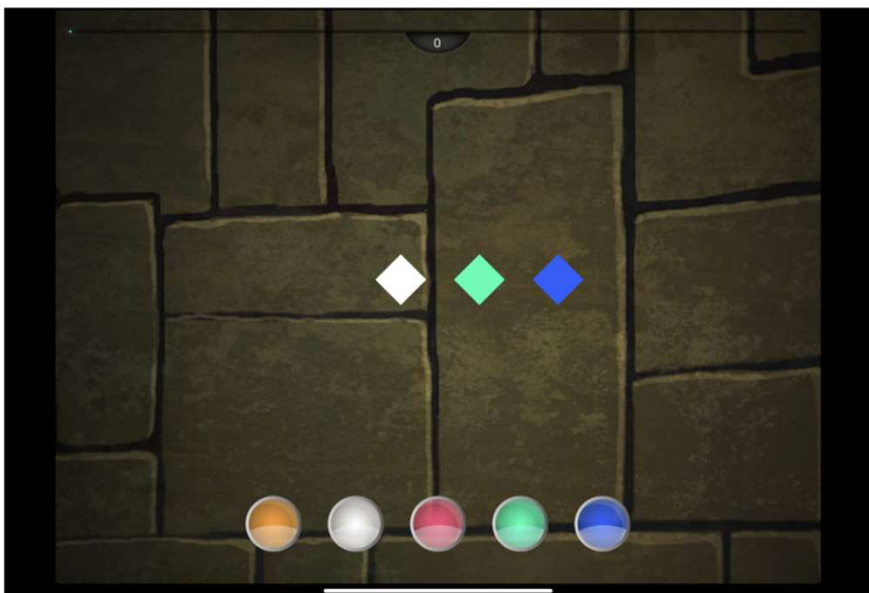




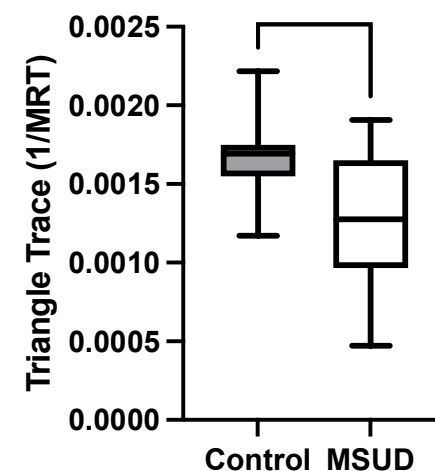
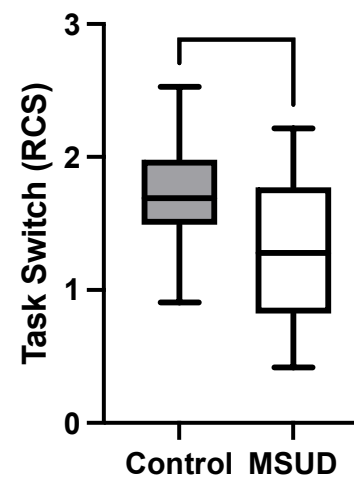
# EF BY WEB-BASED GAME - ATTENTION



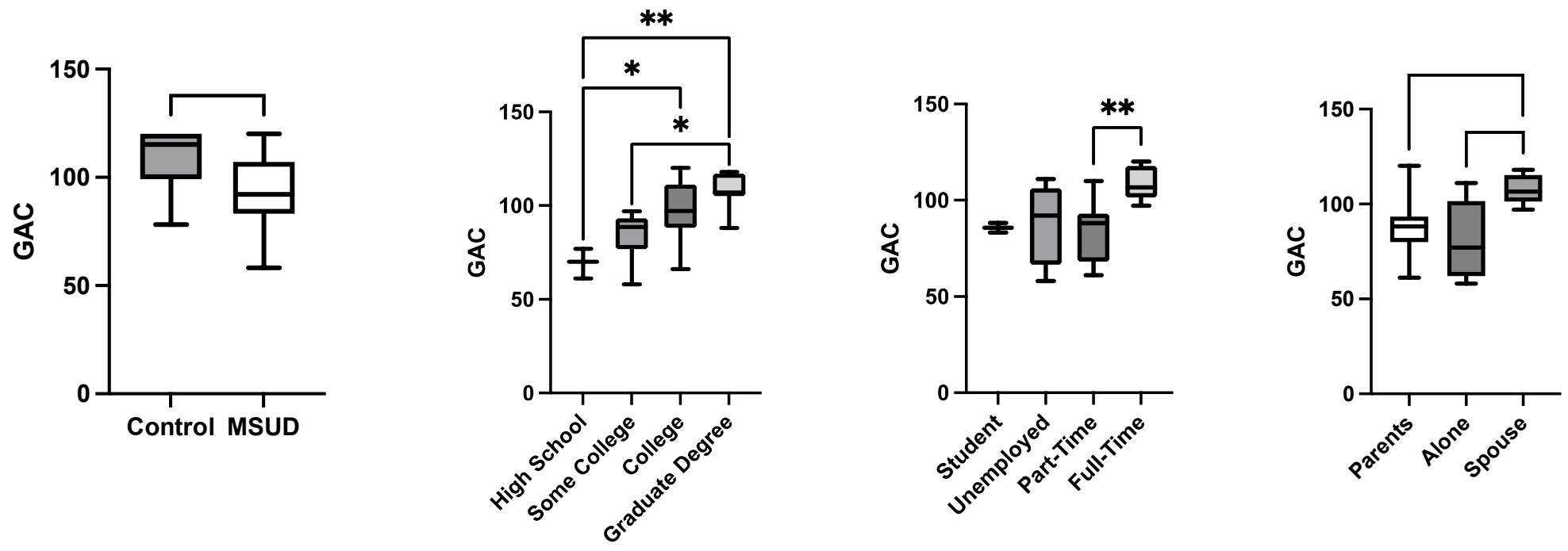
# EF MEASURED BY WEB-BASED GAME - WORKING MEMORY



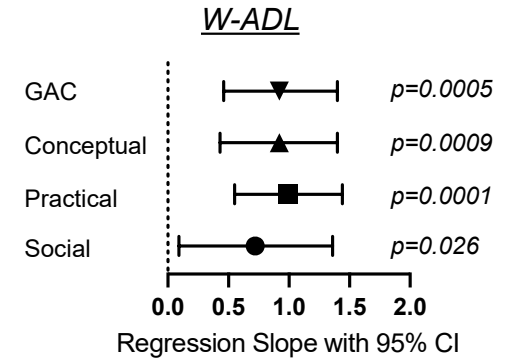
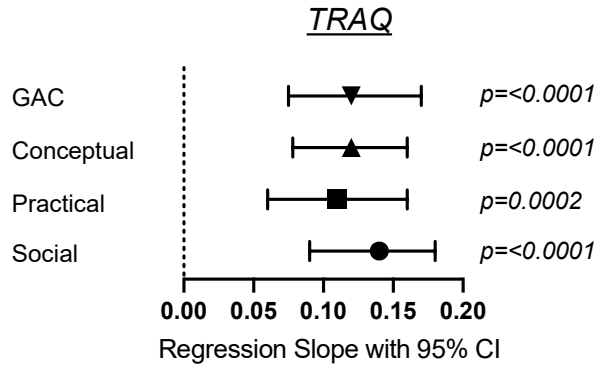
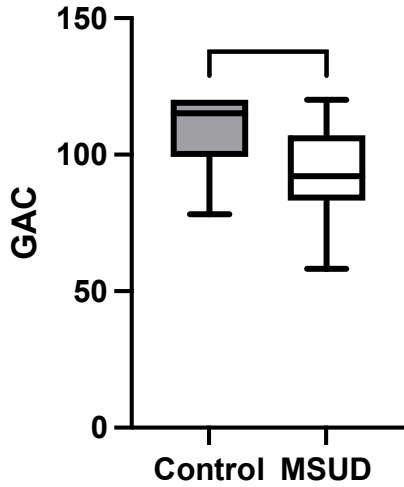
# EF MEASURED BY WEB-BASED GAME - COGNITIVE FLEXIBILITY



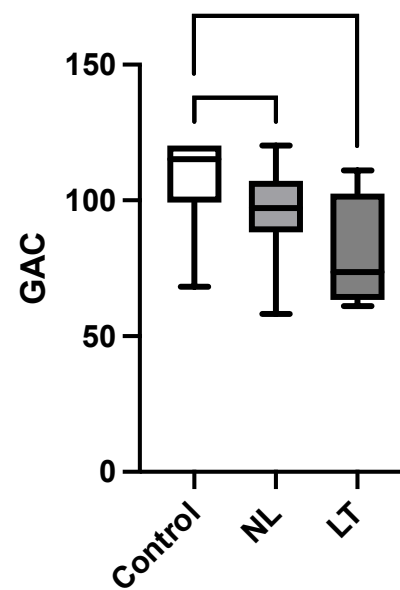
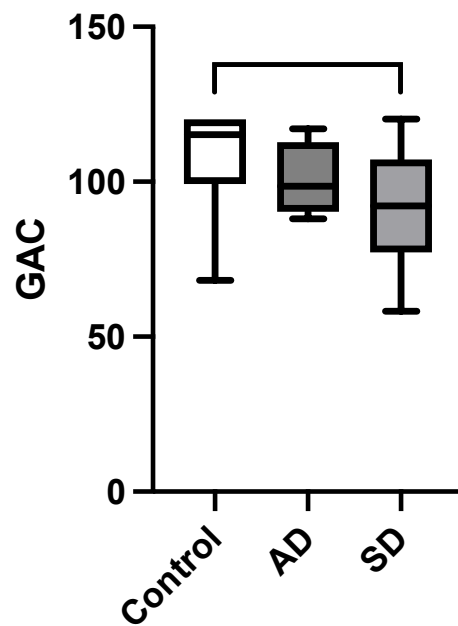
# ABAS-3: ADAPTIVE FUNCTION QUESTIONNAIRE



# ABAS-3: ADAPTIVE FUNCTION QUESTIONNAIRE




# CLINICAL HISTORY MAY IMPACT ABAS-3 SCORES



# THEMES FROM SEMI- STRUCTURED INTERVIEWS

- Diet concerns no longer predominant
  - Affected adults report a turning point when they realized that diet was necessary and increased compliance
- Mental health is a significant concern, especially finding knowledgeable providers
  - Dietary non-adherence is used as an intentional method of self-harm
- Symptoms of hyperleucinoses change in adulthood - more report brain fog and fatigue than ataxia
- Most are satisfied with their metabolic care, even if it is received at pediatric centers
  - There is increased worry about their non-metabolic medical care, including how adult-onset co-morbidities will be managed
- Health care transition routinely addresses MSUD dietary management but does not include education, income, vocational, decision-making and financial supports

# SUMMARY

- Adults with MSUD have significant executive and adaptive function deficits compared to age-matched controls
  - Executive and adaptive function correlated with long-term functional outcomes in education, employment, medical self-management, and independent living skills
  - Adults who were diagnosed asymptotically scored better on executive and adaptive function measurements
  - Adults who received liver transplants score worse on measure of skills for independence
- 



# ACKNOWLEDGEMENTS

Participants and their families

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