Cognitive and Behavioral Profiles in Down Syndrome Across the Life Course

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Cognitive and Behavioral Functioning in Down syndrome (DS)

- Overall, score on IQ measures in mild (IQ: 50-69) to moderate (IQ: 35-49) range (Hamburg et al., 2019)
  - 10-15% severe to profound (IQ > 35) range
  - >5% borderline (IQ: 70-85) or average (IQ: 85+) range
  - full trisomy (mean IQ: 50-55) vs. mosaic (mean IQ: 63-68)
  - Support and education

- At risk for maladaptive behaviors and mental health problems
  - Up to 2/3rds of adults with DS do not qualify for a mental health disorder (Mallard et al., 2014; Mantry et al., 2008)

- Unique profile of strengths and challenges across the life course
Cognition: Infancy and Toddlerhood

Challenges
• Reduced/delayed vocal reactivity and responsiveness
• Delayed development of inhibitory processes
• Delays in canonical babbling and first words
• Expressive language
• Social cognition

Strengths
• Interest in face-to-face interactions
• Joint attention
• Gestural communication
• Language comprehension
• Pragmatic aspects of language
• Visual learning

Maximizing Learning
Understand more than they can say
Allow time to react/respond
Clear, simple phrases and visual cues/modeling
Multiple ways to communicate
Leverage positive interactions
Cognition: Childhood and Adolescence

• **Challenges**
  - Auditory memory
  - Expressive language (esp. syntax, phonological processing and intelligibility)
  - Phonic learning
  - Executive Functions

• **Strengths**
  - Comprehension
  - Social skills
  - Visual learning
  - Visual memory
  - Sight vocabulary
Strength in Visual Learning and Memory

• Strong visuospatial abilities relative to verbal processing skills (Costanzo et al., 2013; Jarrold et al., 1999; Klein & Mervis, 1999; Fidler et al., 2006)

• Aspects of visuospatial processing especially strong (Fidler, 2005)
  • visual memory
  • visual-motor integration
  • visual imitation
**Challenge in Memory & Executive Functions**

### Maximizing Learning

- Direct, clear and short phrases
- Visual demonstrations, pictures, charts
- Learning by doing
- Repetition
- Break tasks into small steps, simplify
- Reduce distraction
- Lots of breaks between periods of learning
- Explicit directions and cues to regulate behavior
Cognition: Adulthood

Challenges
- Auditory memory
- Expressive language skills relative to other adaptive behavior skill areas
- Intelligibility problems and sound production errors
- Executive Functions

Strengths
- Comprehension skills
- Activities of daily living and employment skills
- Visuospatial abilities
- Visual memory and learning
Cognition: Aging in DS

- Alzheimer’s disease
  - Brain pathology - amyloid-beta (Aβ) - present by age 40 years (Fortea et al., 2021; Head et al., 2012; Lao et al., 2017)
  - Over half of adults with DS exhibit Alzheimer’s dementia by age 55 years (Rubenstein et al., 2020)
    - Variability; genes such as APOE (Fortea et al., 2020), health conditions (Lao et al., 2022), and lifestyle (e.g., Mihaila et al., 2020)

- Sequence of cognitive decline
  - Memory & Attention
  - Executive Functions & Visuospatial & visuomotor
  - Language, Gait

Fortea et al., 2020; Lao et al., 2017; Zammit et al., 2020; Head & Lott, 2019

Hartley et al., 2020; Fleming et al., in press
Alzheimer’s Biomarker Consortium - Down Syndrome

This study is for:
- People with Down syndrome
- 25 years of age or older
- Accompanied by study partner (e.g. parent, caregiver, legal guardian)

What does this study involve?
- Up to 4 visits over five years
- Study Partner completes questionnaires
- Participants complete physical exam, blood draw, and tests of thinking
- Participants complete an MRI & PET scans, and an optional lumbar puncture
- Invited to report on lifestyle

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Ways to Preserve Cognitive Functioning with aging in DS

• Sleep
  • Sleep disruptions and sleep disordered breathing associated with executive functioning and memory (Cody et al., 2020; Fleming et al., 2021)
  • Regular sleep screenings and compliance with treatments and recommendations

• Physical Activity
  • More sedentary behavior and too little physical activity associated with poorer executive functioning and memory (Fleming et al., 2021)
  • Build activity into a routine and make social

• Cognitive Stimulation
  • Identify leisure or employment/day program/other activities that make you think (Mihaila et al., 2019)

“…you know the memories might go away, but what will never go away – your family.”

-Danny Chafetz, Edgewood College Cutting Edge Program
Profile of Maladaptive Behaviors - Childhood/Adolescence

Challenges- Toddlers & Children
- Wondering
- Disruptive and impulsive and hyperactive
- Oppositional and disruptive behavior
- Anxious, inflexible and stuck behaviors

Challenges- Adolescence
- Depression, social withdraw
- Generalized anxiety
- Obsessive compulsive behaviors

Strengths
- Social motivation
- Routines
- Social skills
Autism

- 5-16% children with DS have diagnosis of autism spectrum disorder (Richards et al., 2018)

- Frequency and severity of autism symptoms negatively associated with IQ and adaptive functioning (cite)

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Autism
- Insistence on routines/sameness
- Limited expressive language
- Repetitive play
- Delayed development
- Anxiety
- Poor perspective taking i& social cognition
- Reduced interest in communicating
- Reduced use of gestures
- Little non-verbal communication
- Can exhibit social indifference
- May avoidance of eye contact

Down syndrome
- Interested in people
- Eye contact
- Joint attention
- Symbolic play
- Imitate others
- Language develops normal sequence

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Profiles of Maladaptive Behavior - Adults

Challenges
• Regression
• Anxiety and depression
• Obsessive compulsive behavior
• Self-Talk

Strengths
• Employment and daily living skills
• Routines
• Social skills

Self-Talk
91% of children and adults engage in self-talk
(Patti, Andloro, & Gavin, 2008)

Employment
- Janitorial Work
- Clerical work
- Speaking/Advocacy
- Food industry
- Bagging/shelving
  - hosting
  - assembly
  - College
Regression

- Sudden or progressive ‘regression’
- 1-17%; late adolescence to early adulthood
- Range of symptoms – changes in social skills, adaptive behavior, attention, and internalizing behavior most common (see Rosso et al., 2019; Walpert et al., 2021)
- Treatment often anti-depressants or anti-psychotics
  - Typically partial recovery of abilities (Walpert et al., 2021)
- Not Alzheimer’s disease, but could share etiological mechanisms and/or alter timing (i.e., tau and NFL) (Handen et al., 2021)
Summary

- Individuals with DS have a unique profile of strengths and challenges in cognition and behavior.

- Leverage strengths and reduce impact of challenges to promote learning and enhance quality of life.

- Profiles presented on today are based on average patterns; there is a TON of variable among children and adults with DS. Many individuals with DS will have additional or alternative strengths or challenges.
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