

Down Syndrome Fragile X Syndrome Fetal Alcohol Syndrome

Project ECHO

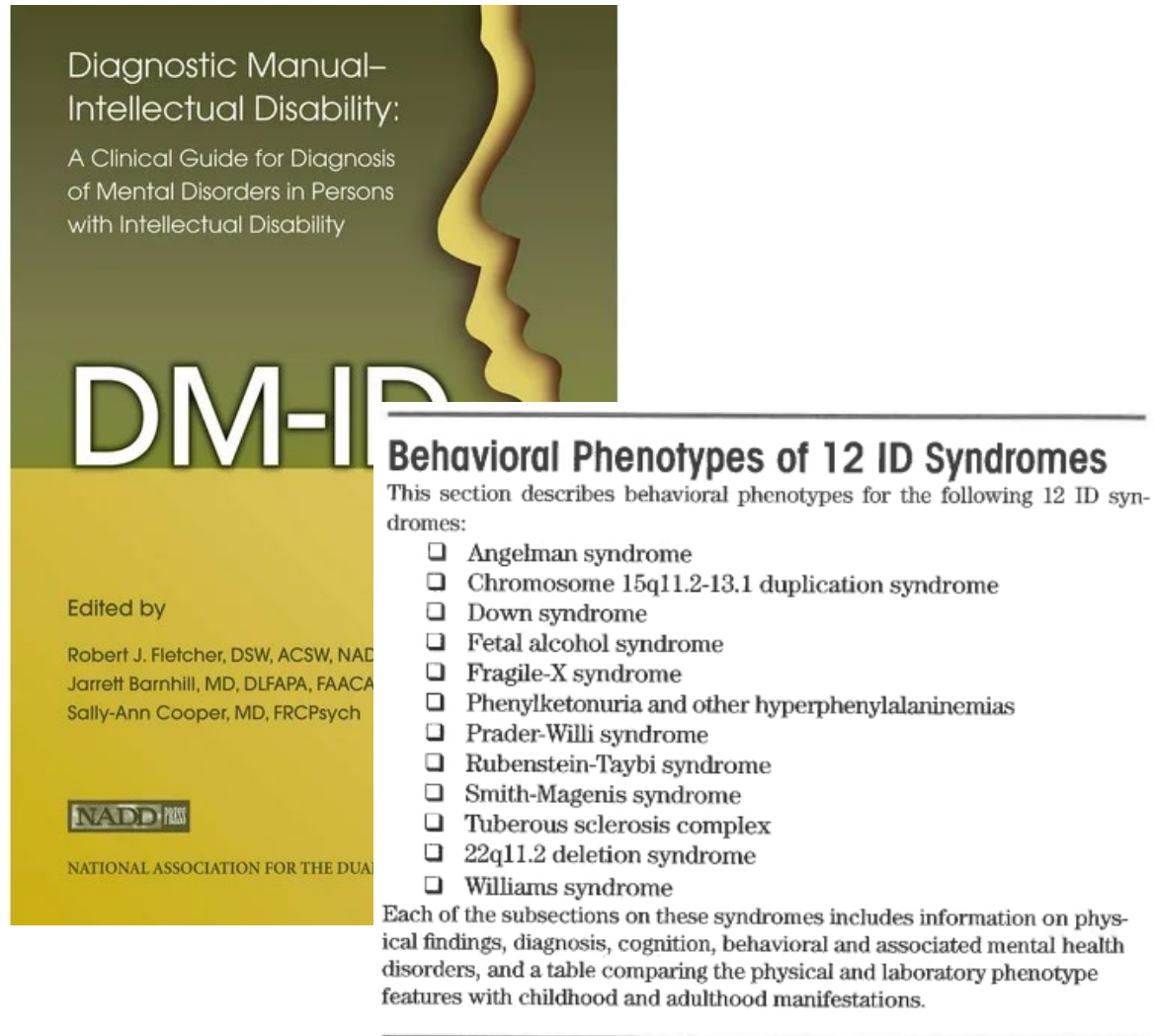
7/23/2026

Jodi Tate, MD

Overview

- 12 syndromes associated with ID that have known phenotypes
- Importance of knowing if your client/patient has one of these syndromes
- 3 most common causes of ID
 - Down Syndrome
 - Fragile X Syndrome
 - Fetal Alcohol Syndrome

12 ID syndromes with known phenotypes



Definitions

- Physical phenotype
 - Common physical characteristics
- Behavioral phenotype
 - Common behavioral/mental health characteristics

Why is it important
to know if your
client/patient has
one of these
syndromes?

Improves Medical Care

Helps predict future needs

Guides educational/behavioral
supports

Provides information for family
members

Connect people with resources and
community

Health Watch Tables!

[Health Watch Tables - Health Care for Adults with Intellectual and Developmental Disabilities](#)

Health Watch Tables

Health Watch Tables for Selected Developmental and Related Disabilities

Health Watch Tables provide health information for specific syndromes with an established etiology and with identified health concerns. These syndrome specific tables were developed to complement the 2011 version of the [Canadian consensus guidelines on the primary care of adults with developmental disabilities](#) and the Preventive Care Checklist for adults with intellectual and developmental disabilities.

Recommend:

Print off and bring to your client's healthcare appointments

[Autism Spectrum Disorder](#)

[Down Syndrome](#)

[Fetal Alcohol Spectrum Disorder](#)

[Fragile X Syndrome](#)

[Prader-Willi Syndrome](#)

[Williams Syndrome](#)

[22q11.2 Deletion Syndrome](#)

3 most common causes of ID

Down syndrome

*most common chromosomal cause of ID

Fragile X syndrome

*most common inherited genetic cause of ID

Fetal Alcohol Spectrum Disorders

*most common preventable cause of ID

Down syndrome

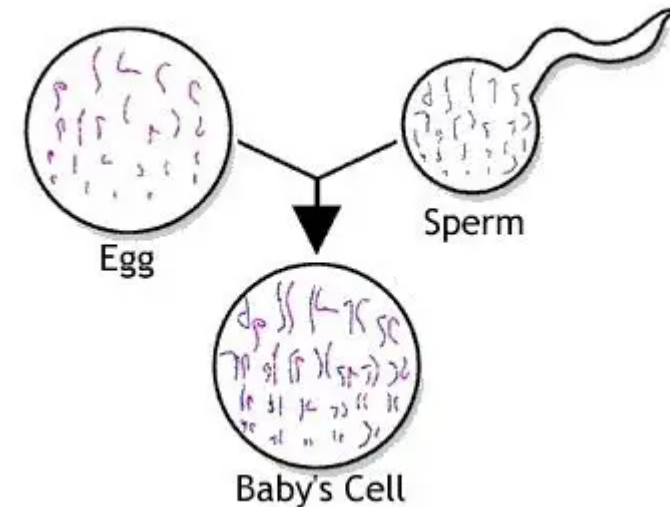
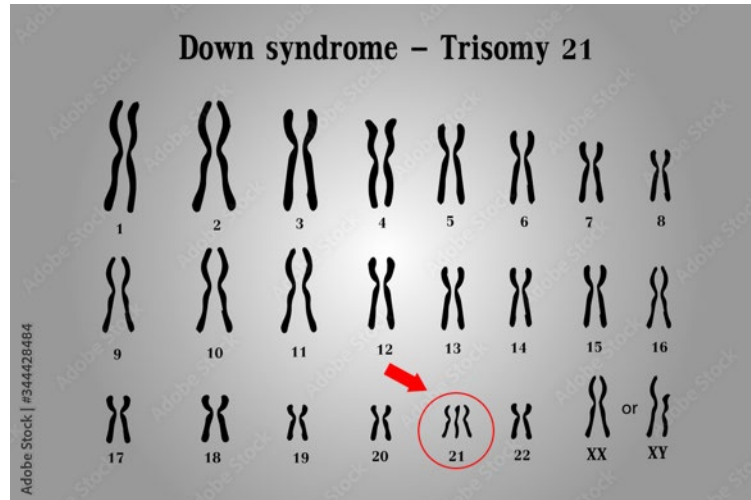
*most common chromosomal cause of ID

- Down Syndrome - Health Care for Adults with Intellectual and Developmental Disabilities

Health Care for Adults with Intellectual and Developmental Disabilities	
Down Syndrome	
<i>Health Watch Table</i>	
1. Head, eyes, ears, nose, throat	
Considerations	Recommendations

What is the cause of Down Syndrome?

- Down Syndrome
 - Extra copy of chromosome 21
 - Total of 47 chromosomes
 - Trisomy 21
- Human chromosomes (karyotype)
 - 46 chromosomes
 - 23 from sperm
 - 23 from egg



What are common physical characteristics?



Common Medical Conditions

- Hearing Loss
- Heart defects
- Obstructive Sleep Apnea
- Hypothyroidism
- Cancer
- Immune System
- Feeding difficulties/GI

- Check out the LowDOWN Podcast

[John's Crazy Socks - Down Syndrome Resource Foundation](#)

What are common behavioral/mental health characteristics?

- Self Talk is very common
 - Common and developmentally appropriate
 - Self talk can increase in times of stress
 - Often mistaken for psychosis
- Strong willed
- ADHD
- Autism
- Depression/Anxiety
- Obsessive Compulsive Disorder
- Dementia
- Psychosis is rare/atypical



Fragile X syndrome

*most common inherited genetic cause of ID

- [Fragile X Syndrome - Health Care for Adults with Intellectual and Developmental Disabilities](#)

Health Care for Adults with Intellectual and Developmental Disabilities

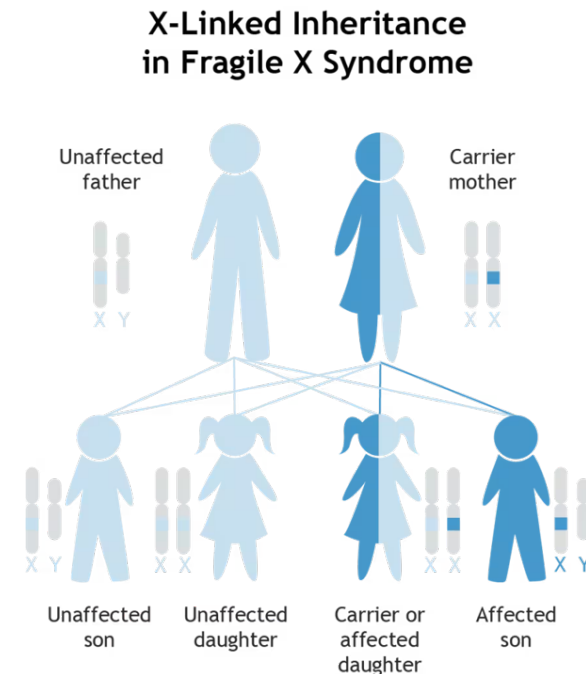
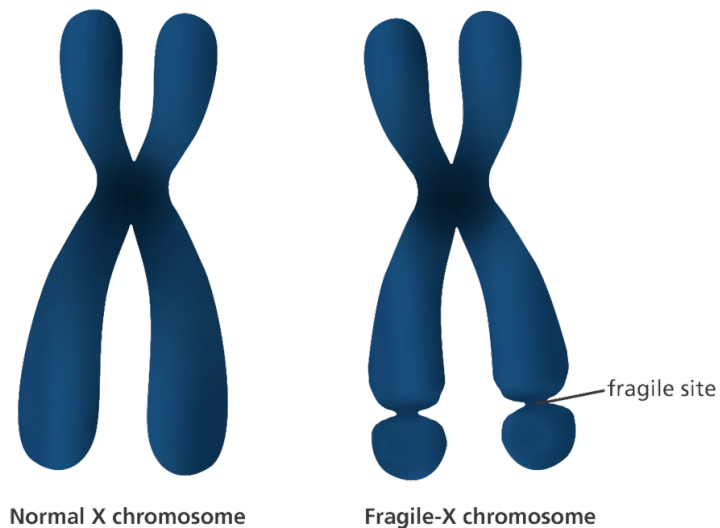
Fragile X Syndrome

Health Watch Table

1. Head, eyes, ears, nose, throat

What is the cause of Fragile X Syndrome?

- Cause by a mutation of the FMR1 (Fragile X Mental Retardation) gene on the X chromosome
- Discovered in 1991
- Males: XY Females: XX
- Males are more severity affected as they only have one x chromosome



What are common physical characteristics?



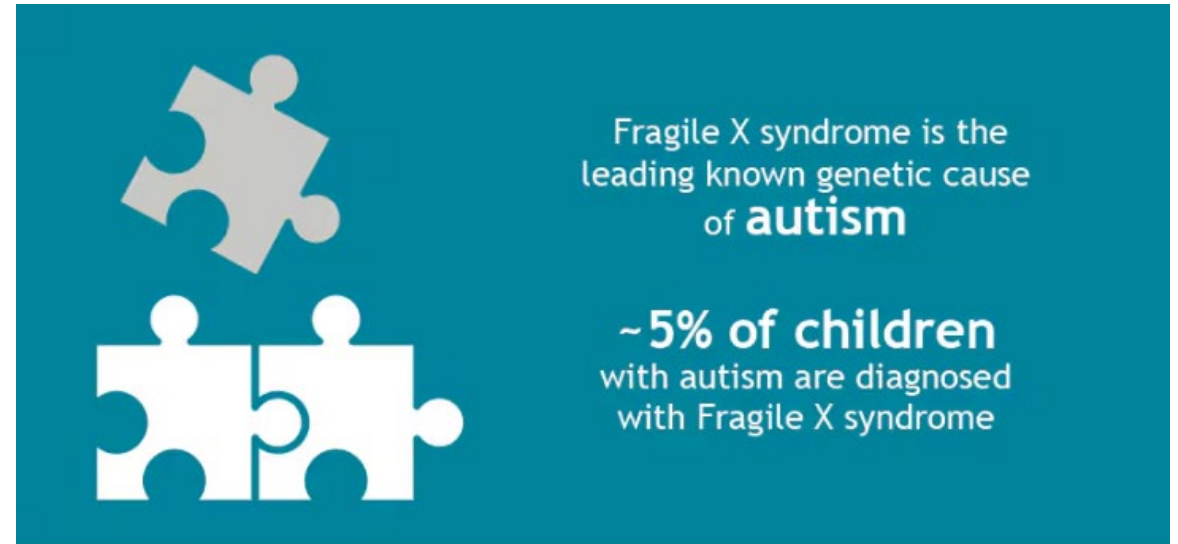
Common Medical Conditions

- Heart valve
- Seizures
- Ear infections
- Sleep/Obstructive Sleep Apnea
- Hernia/Large testicles
- Low muscle tone/Flat feet
- Flat feet
- Vision/Hearing

<https://positiveexposure.org/album/fragile-x-syndrome/>

What are common behavioral/mental health characteristics?

- Males more severity affected than females
- Borderline to mild ID
- Stereotype and repetitive behaviors (hand flapping)
- **Autism Spectrum Disorders**
- Mood/Anxiety
- ADHD



<https://asuragen.com/discoverfragilex/prospective-parents/>

Fetal Alcohol Spectrum Disorders

*most common preventable cause of ID

- Fetal Alcohol Spectrum Disorder (FASD) - Health Care for Adults with Intellectual and Developmental Disabilities

Health Care for Adults with Intellectual and Developmental Disabilities

Fetal Alcohol Spectrum Disorder (FASD)

Health Watch Table

Tao, Temple, Casson and Kirkpatrick 2013

Overview: Fetal Alcohol Spectrum Disorder is an umbrella term for the range of effects that can occur in an individual exposed to alcohol in utero. These effects can include various physical, intellectual and neurobehavioral deficits that vary widely in severity. Fetal alcohol syndrome (FAS), partial fetal alcohol syndrome (pFAS), alcohol-related neurodevelopmental disorder (ARND) and alcohol-related birth defects (ARBD) are now used to refer to each of the four sub-categories subsumed under FASD.^{1,2} Prenatal alcohol exposure does not always lead to FASD.

Prevalence: The reported incidence of full FAS currently ranges from 0.2 to 2.0 cases per 1,000 live births and up to 43 per 1,000 among “heavy” drinkers (different population surveyed or different methods used). The prevalence of FASD has been estimated to be at 9.1 per 1,000 live births in the United States.^{3,4}

Etiology: Prenatal alcohol exposure

The range of deficits in FASD is associated with many factors, including the amount, time and frequency of exposure, as well as the state of health/nutrition of the mother and the genetic makeup of the mother and the fetus. ***There is no time during pregnancy when it is shown to be safe for mothers to consume alcohol. While binge drinking is especially dangerous, no amount of alcohol has been shown to be safe for all women.***

1. Diagnosis



Fetal Alcohol Spectrum Disorders



What Are the Types of Fetal Alcohol Spectrum Disorders?

The term *fetal alcohol spectrum disorders* (FASD) refers to a range of physical, cognitive, and behavioral abnormalities caused by prenatal alcohol exposure. Depending on the features identified, the disorders categorized as FASD^{15,16} include:

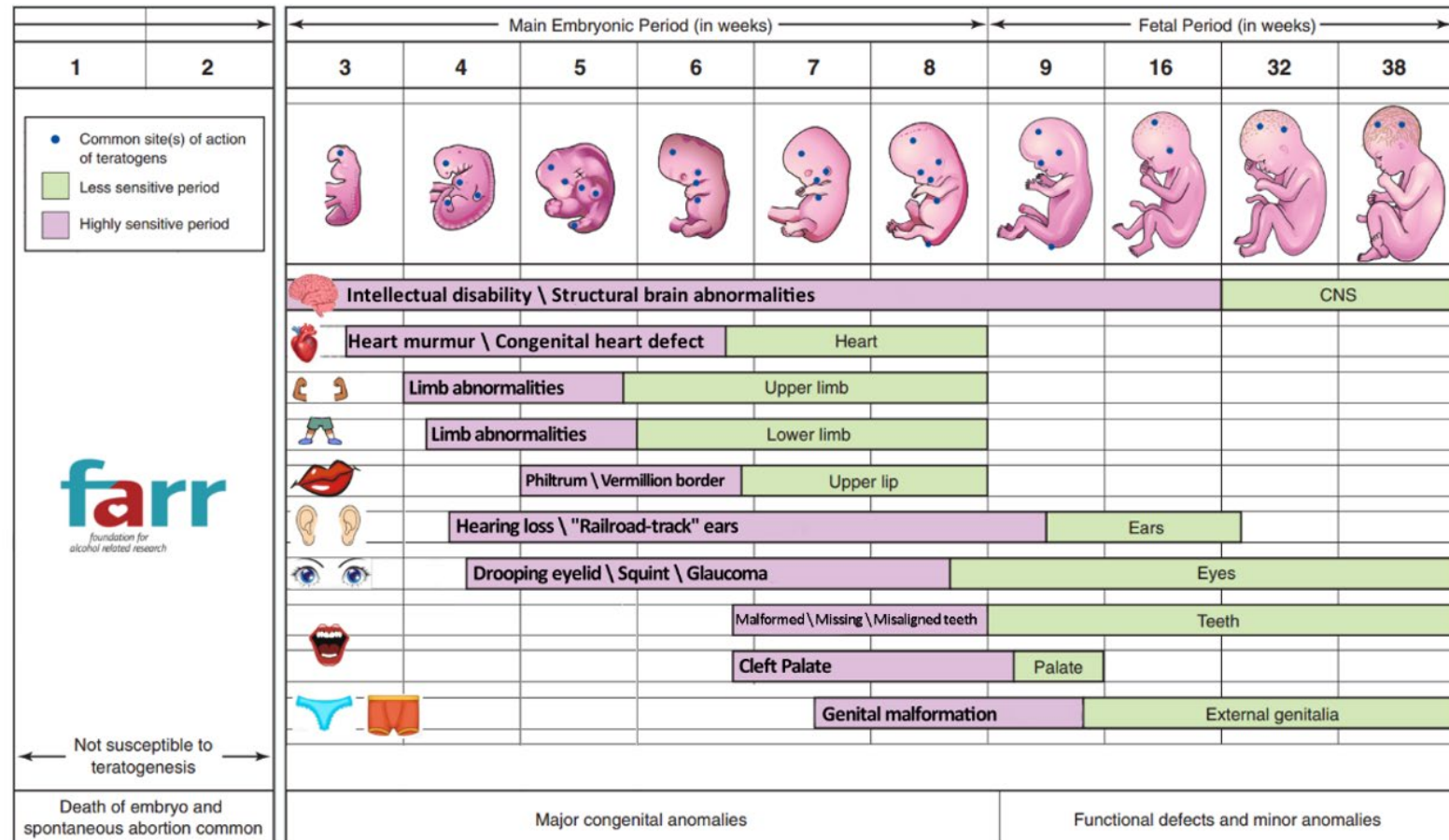
- Fetal alcohol syndrome
- Partial fetal alcohol syndrome
- Alcohol-related neurodevelopmental disorder
- Alcohol-related birth defects
- Neurobehavioral disorder associated with prenatal alcohol exposure

Prenatal alcohol exposure and central nervous system (CNS) involvement are factors common to the disorders encompassing FASD. Evidence of CNS involvement can be structural (e.g., small brain size, alterations in specific brain regions) or functional (e.g., cognitive and behavioral deficits, motor and coordination problems). For fetal alcohol syndrome—which is also characterized by growth deficiencies, distinct facial features, and other physical factors in addition to CNS involvement—confirmation of prenatal alcohol exposure is not required.

What is the cause of Fetal Alcohol Spectrum Disorders?



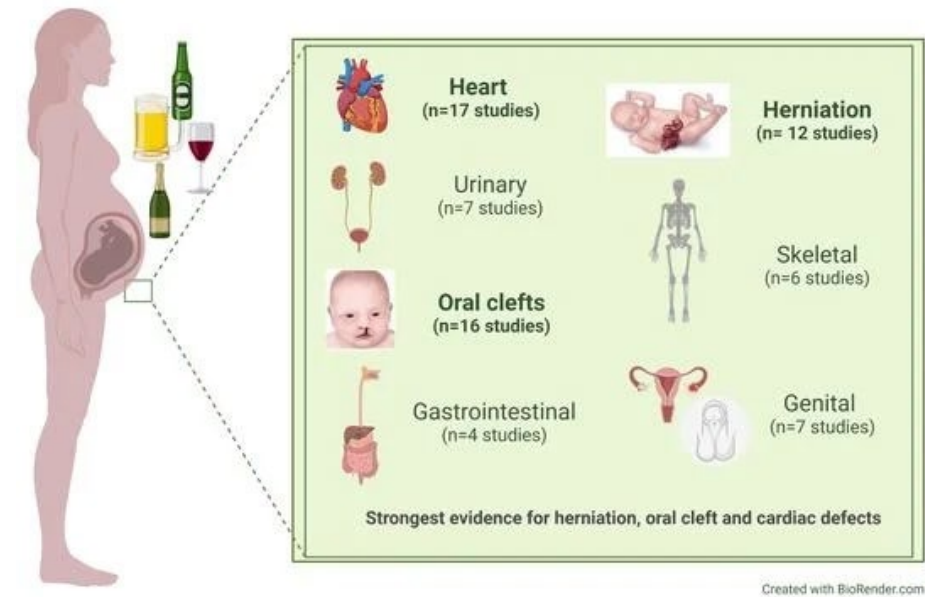
Here's how alcohol affects an unborn baby at each stage of pregnancy



Adapted from: Moore, K. L., Persaud, T. V. N., & Torchia, M. G. (2016). Before we are born: essentials of embryology and birth defects (9th ed.). <http://doi.org/9780323313407>

[International Fetal Alcohol Syndrome day - 9 September 2020 - Optima](#)

What are common physical characteristics?



Dyląg, (2023). Birth Defects Associated with Prenatal Alcohol Exposure—A Review. *Children*, 10(5), 811. <https://doi.org/10.3390/children10050811>

Specific facial characteristic that are hard to recognize

What are common behavioral characteristics?

- Range of intellectual deficits
- Executive functioning deficits
- Social skills deficits
- Mood and anxiety disorders
- ADHD
- Personality disorders
- Substance use disorders

Ten Brain Domains (Functions) Affected by FASD

Alcohol affects the growth and formation of the brain, and this is often seen in an individual's behavior and development. The following brain functions or domains are evaluated by a diagnostic team during an FASD assessment.

ACADEMIC ACHIEVEMENT

- May have difficulty in school: reading, math, comprehension (understanding) and abstract concepts

ATTENTION

- Can be easily distracted, difficulty paying attention and sitting still

COGNITION

- Difficulty reasoning, planning, solving problems and understanding complex ideas
- Wide range of IQ scores are found

LANGUAGE (Expressive and receptive)

- Delay in language development
- Difficulty understanding lengthy conversation and instructions
- May speak well, but not fully grasp the meaning
- Can repeat instructions or rules, but may not follow through

MEMORY

- Difficulty with long-term, short-term and working memory
- May appear to lie, but is actually filling in the blanks when unable to remember
- Trouble with memorizing and may seem forgetful
- Difficulty with accessing, selecting and organizing information when needed

NEUROANATOMY/NEUROPHYSIOLOGY (Brain structure and function)

- Could have a smaller head, brain size, seizure disorder and/or abnormal findings on a scan (ex: MRI or EEG) consistent with prenatal alcohol exposure

EXECUTIVE FUNCTIONING

- May have trouble with planning, sequencing, problem solving and organizing
- May be impulsive and/or hyperactive
- Difficulty understanding cause and effect and controlling behaviour
- Challenges with transitions and change
- Often repeats mistakes
- Difficulty with concepts, abstracts ideas, consequences and managing time

ADAPTIVE BEHAVIOR, SOCIAL SKILLS AND SOCIAL COMMUNICATION

- May not understand personal boundaries and have difficulty reading social cues
- May be socially vulnerable and easily taken advantage of
- Difficulty seeing things from another's perspective
- Socially and emotionally immature and may behave younger than actual age
- May have trouble with hygiene, money and coping skills

MOTOR SKILLS

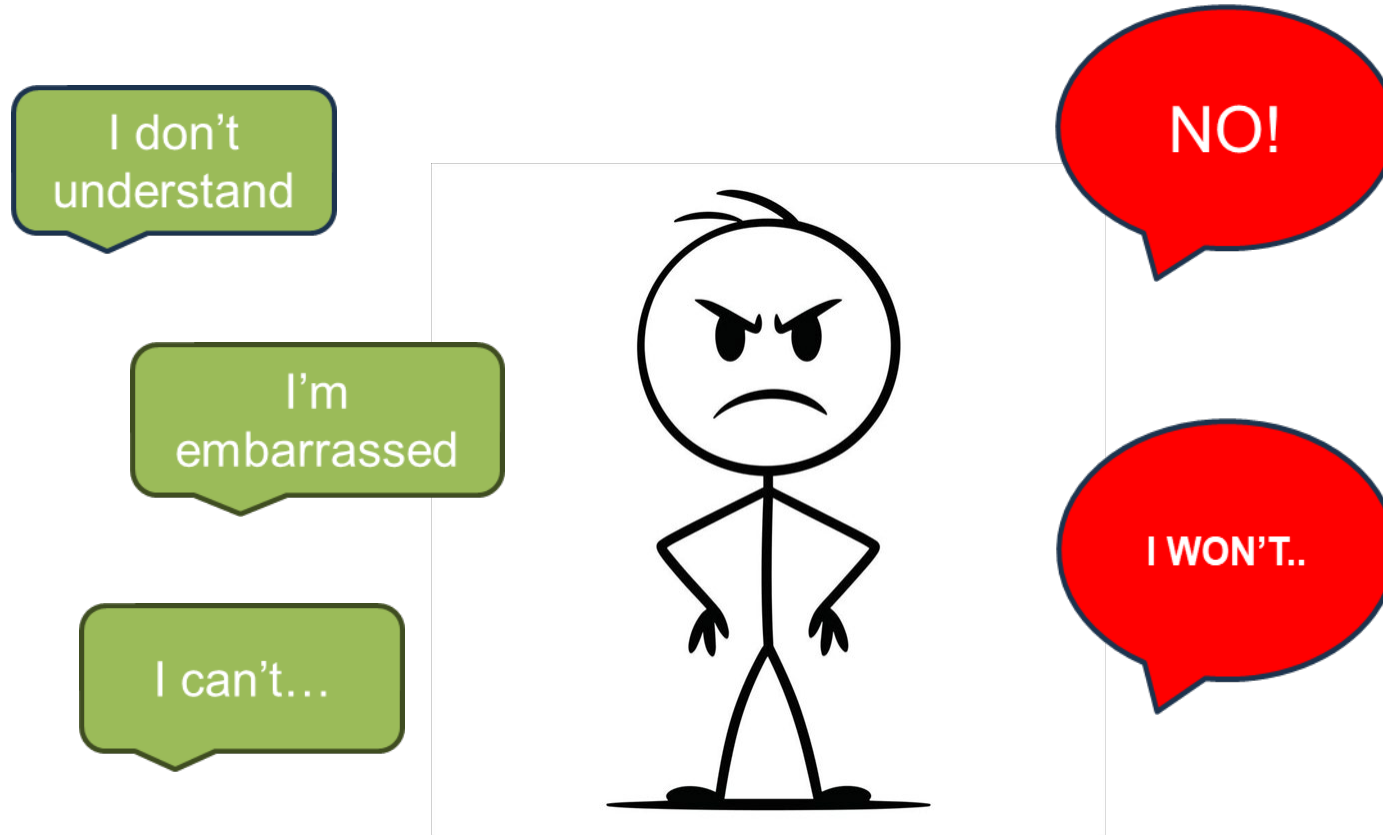
- Difficulty with balance, strength, endurance, coordination, reflexes and muscle tone
- Difficulty with printing, using pencil and scissors

AFFECT REGULATION

- Includes anxiety, depression and mood imbalance in the severe range meets Diagnostic and Statistical Manual-V criteria

The Canadian FASD Diagnostic Guidelines were revised in 2015 and the Sensory Domain has been removed from the list above; however, the Manitoba FASD Centre will continue to measure and consider sensory functioning (see below) during an FASD assessment.

Important to reframing perceptions



Reframing Perceptions From "Won't" to "Can't" (Diane Malbin)

The shift is from seeing a child as one who **won't** do something, to one who possibly **can't**.

The shift includes moving from:

FROM SEEING CHILD AS:

won't
bad
lazy
lies
doesn't try
mean
doesn't care, shut down
refuses to sit still
fussy, demanding
resisting
trying to make me mad
trying to get attention
acting younger
thief
doesn't try
inappropriate
not trying to get the obvious

PERSONAL SHIFT FROM:

hopelessness
fear
chaos, confusion
anger
power struggles
frustration
exhaustion
no good outcomes
isolation

PROFESSIONAL SHIFT FROM:

traditional
applying consequences
traditional interventions
changing people

TO UNDERSTANDING CHILD AS:

can't
frustrated, defended, challenged
tries hard
confabulates/ fills in
exhausted or can't start
defensive, hurt, abused
can't show feelings
overstimulated
oversensitive
doesn't get it
can't remember
needing contact, support
being younger
doesn't understand ownership
tired of always failing
may not understand proprieties
needing many reteachings

TO FEELINGS OF:

hope
understanding
organization, meaningfulness
reframing perceptions, defusing
working with, rather than at
trying differently, not harder
re-energized, new options to try
seeing, supporting strengths
networking, collaboration

TO:

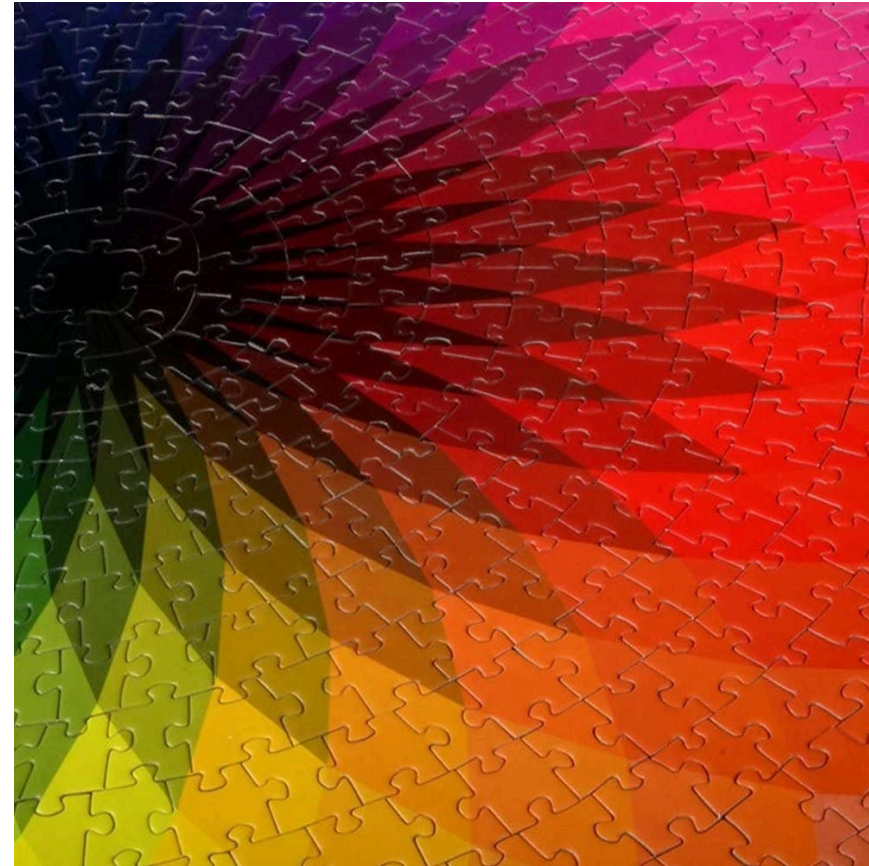
recognizing brain differences
preventing problems
expanding professional options,
develop effective strategies
changing environments

*Source: Copyright © 2002, 1999,
Diane V. Malbin.
Used with permission.

Behavioral Symptoms & Accommodations for Fetal Alcohol Spectrum Disorder (FASD)			
Behavioral Symptoms:	Can look like:	Brain differences	Accommodations
Saying "no" or refusing requests, asking the same question over and over, saying "huh?" or "what?" a lot, not acting when requests are made	Not listening, ignoring, defiance, opposition, daydreaming.	Slow auditory processing	Slow down. Talk less. Give time. Provide visuals, or lists. Teach using hands-on and kinesthetic methods.
Socially and emotionally like a younger child, interest in activities similar to that of a younger person, unable to "act their age," overly friendly with people, misunderstanding personal boundaries	Immaturity, babyishness, laziness, irresponsibility, disregard for consequences, purposefully irritating, pushy.	Dysmaturity, or developmental delay. Poor adaptive functioning	Recognize developmental age (vs. chronological) Adjust expectations to match developmental age Provide opportunities for friendships/social situations that match person's developmental age vs. their chronological.
Difficulty remembering information learned recently, difficulty remembering multiple instructions, may remember one day, not another, repeatedly making the same mistakes. Confabulation.	Manipulation, not trying, purposeful misbehavior, "doesn't care," disrespect, lying.	Impaired memory	Repeat, remind, and provide visual and mnemonic clues. Recognize need to reteach. Drop judgment. Understand confabulation. Don't punish.
Difficulty waiting turn, difficulty complying with rules, interrupting, "see it-want it-take it," blurting, inappropriate language, risk taking	Rudeness, opposition, lying, disrespect, danger seeking.	Impulsivity	Provide structure and support. Understand. Provide non-verbal reminders
Difficulty solving problems, poor organization, trouble making a plan, difficulty predicting outcomes, difficulty with organization, perseveration, inflexibility.	Stubbornness, laziness, "doesn't care" about consequences, messy, disrespect, badgering.	Poor executive functioning	Provide structure. Provide organizational help. Support, support, support! Some people call this an "External Brain."
Frequent behavioral outbursts, low mood, moodiness, explosive reactions, fight or flight, anxiety.	Aggression, anger, opposition, disrespect, depression.	Difficulty modulating emotions	Look for triggers to prevent problems. Anxiety can look like anger or irritability. Allow breaks Figure out what helps soothe and reset, and provide opportunities for this.
Over responsive to stimuli and under responsive to stimuli, often both in the same person, hyperactivity, distractibility, inattention, social difficulties, learning difficulties, emotional reactivity, clumsiness, and poor organizational skills.	Irritability, opposition, "pickiness," manipulation, tantruming, rule breaking, anxiety, aggression, avoidance.	Sensory differences	Look for triggers. Remove or avoid to prevent problems. Figure out what helps soothe and reset, and provide opportunities for this. For those sensory differences that get in the way of safety or healthy living (sensory seeking behaviors, lower/no sensation of heat/cold, lower/no sensation of hunger) provide structure and support.

Summary and Thank you!

- Important to know if your client has a syndrome that is associated with ID
- Be familiar with the health watch tables to advocate for your client
 - [Health Watch Tables - Health Care for Adults with Intellectual and Developmental Disabilities](#)
- Syndrome Sheets is another resource
 - [Syndrome Sheets – SSBP](#)



Down Syndrome

Down Syndrome

Etiology: Trisomy 21 (extra chromosome 21)

[Down Syndrome - Health Care for Adults with Intellectual and Developmental Disabilities](#)

Physical Phenotype

- Small head, mouth
- Upslant to eyes
- Epicanthal folds
- Broad neck
- Hypothyroidism
- Hearing loss/visual impairments
- Cardiac problems
- Gastrointestinal
- Orthopedic, skin
- Obesity

Behavior Phenotype

- Self-talk
- Strong willed
- ADHD
- Social Anxiety
- Depression/Anxiety
- OCD
- Atypical psychosis



Fragile X Syndrome

Fragile X Syndrome (FXS)

Etiology: mutation in the FMR1 gene, located at Xq27.3

Fragile X Syndrome - Health Care for Adults with Intellectual and Developmental Disabilities

Physical Phenotype

- Visual impairments
- Hearing impairments
- Recurrent ear infections
- Seizure disorder
- Aortic dilation
- Connective tissue anomalies
- Hernia
- Macroorchidism (large testicles)
- Long face

Behavior Phenotype

- Autism Spectrum Disorders
- Borderline to mild ID (males more severely affected)
- Stereotypy and repetitive behaviors (hand flapping)
- Mood and anxiety disorders
- ADHD
- Fragile X Tremor Associated Syndrome (FXTAS)

Fetal Alcohol Syndrome

Fetal Alcohol Spectrum Disorder (FASD)

Etiology: exposure to alcohol in-utero

Fetal Alcohol Spectrum Disorder (FASD) - Health Care for Adults with Intellectual and Developmental Disabilities

Physical Phenotype

- Growth deficits
- Cardiac abnormality
- Skeletal abnormalities

Behavior Phenotype

- Range of intellectual deficits
- Executive functioning deficits
- Social skills deficits
- Mood and anxiety disorders
- ADHD
- Personality disorders
- Substance use disorders