

DOWN SYNDROME



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WHAT IS IT?

- Down Syndrome is a genetic disorder in which the majority of kids have an extra full or partial copy of a chromosome on the 21st pair resulting in trisomy 21. A smaller percentage have a mosaic (some cells have an extra copy) or a translocation (part of 21 breaks off and attaches to another chromosome).

PRESENTATION

- Intellectual disability
- Developmental delay
- Congenital heart defects
- Atlanto-axial instability
- Flattened face
- Small head and short neck
- Protruding tongue
- Upward slanted eyelids
- Broad hands with one crease in palms
- Excessive flexibility
- Tiny white spots on iris
- Short height
- Low muscle tone

SPECIAL TESTS



- Pre-natal Screening - can indicate likelihood that mother is carrying a child with DS but it cannot diagnose
 - Blood tests
 - Nuchal translucency test
 - Integrated screening tests
- Pre-natal diagnostic tests
 - Chorionic Villus Sampling
 - Amniocentesis
- Will do a chromosomal karyotype to diagnose

DIFFERENTIAL DIAGNOSIS

- Trisomy 18
- Multiple X Chromosome
- Zellweger Syndrome
- Other peroxisomal problems



TREATMENT EXAMPLES



- Strengthening and postural control, coordination activities
- Education to prevent damage to ligamentous laxity
- Regular exercise to prevent obesity due to increased preference for sedentary activities
- Orthotic recommendations

EXAMPLE QUESTION

Which of the following is not a typical presentation of a child with Down syndrome?

1. Hypertonia
2. Ligamentous laxity
3. Delayed acquisition of motor skills
4. Excessive flexibility

Answer: hypertonia

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