

Fast Facts

Down Syndrome

- Genetic disorder caused when there is abnormal cell division of the 21st chromosome resulting in it getting an extra full or partial chromosome
- Three types: *Trisomy 21* (most common at 95%) is when a person has three copies of chromosome 21 (instead of the normal two). *Mosaic* is a rare form and is when the affected individual only has some cells in their body with an extra copy of chromosome 21. *Translocation* is when a portion of an extra chromosome 21 becomes attached (translocated) to another chromosome other than chromosome 21 (these individuals will have two copies of chromosome 21 and the extra the material on another chromosome).
- Risk factors include being a carrier of translocated genes (can come from mother or father), have one child with Down Syndrome and the age of the mother being over 35 (older eggs can have a greater chance of not being able to properly divided chromosomes).

<i>Symptoms</i>	<i>Treatment</i>	<i>Prevention</i>
<ul style="list-style-type: none"> -Flattened face -Small head -Short neck -Protruding tongue -Unusually shaped/small ears -Upward slanting eye lids (called palpebral fissures) -Poor muscle tone -Broad/short hands with a single crease in the palm -Short fingers -Small hands/feet -Excessive flexibility -Short stature -Tiny white spots on iris (colored part of your eye), called Brushfield's spots. -Mild to moderate cognitive impairment -Speech delays -Short-term and long-term memory problems -Other medical conditions: heart problems, obesity gastrointestinal problems, immune disorders, spinal problems, sleep apnea, dementia and leukemia 	<ul style="list-style-type: none"> -Early intervention through early diagnosis either during pregnancy or shortly after birth -Assemble a team of professionals that provide the best treatment possible to the individual's particular needs—can include: primary pediatrician, pediatric cardiologist, pediatric gastroenterologist, pediatric endocrinologist, pediatric neurologist, developmental pediatrician, pediatric ENT (ears, nose and throat doctor), pediatric ophthalmologist, audiologist, speech pathologist, physical therapist and/or occupational therapist -Make use of early intervention programs -Learn about educational opportunities -Find families going through a similar situation to have a support group -Include individual in social and leisure activities (adapt as needed) -Encourage independence -Plan for a transition to adulthood 	None

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