

Down Syndrome: A Parent's Guide



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What is Down Syndrome?

Down Syndrome, also known as Trisomy 21, is a genetic condition that occurs when an individual has an extra copy of chromosome 21.



Types of Down Syndrome:

There are three main types of Down Syndrome:

Trisomy 21:

This is the most common type, where an extra chromosome 21 is present in all cells.

Translocation Down Syndrome:

In this type, a part of chromosome 21 breaks off and attaches to another chromosome. It accounts for a smaller percentage of cases.

Mosaic Down Syndrome:

In this rare form, the extra chromosome 21 is not present in all cells, only in a portion.



Checklist for Parents: Characteristics of Down Syndrome:

Here's a checklist of common characteristics associated with Down Syndrome:

- ✓ Intellectual and developmental delays
- Slanted eyes
- Flattened facial profile
- ✓ Short neck
- ✓ Short stature
- √ Joint hypermobility
- ✓ Low muscle tone (hypotonia)
- Small hands and feet
- Single palmar crease (a single line across the palm)
- ✓ Unique fingerprint patterns
- ✓ Heart murmurs & digestive issues (not in all cases)
- ✓ Hearing or vision problems
- ✓ Delayed speech and language development



How is Down Syndrome Diagnosed?

Down Syndrome can be diagnosed through various methods, including:

Prenatal Screening Tests: These include blood tests and ultrasound scans during pregnancy to estimate the risk.

Diagnostic Tests: Amniocentesis and chorionic villus sampling (CVS) can confirm the diagnosis by analysing the baby's chromosomes.

Postnatal Clinical Assessment: A doctor can diagnose Down Syndrome based on physical characteristics and genetic testing.





Who Diagnoses Down Syndrome?

Medical professionals such as Obstetricians, Genetic Counsellors, and Paediatricians are typically involved in diagnosing Down Syndrome.



If You Suspect Your Child Has Down Syndrome:

If you suspect that your child has Down Syndrome, here are the steps you can take:

1. Consult a Paediatrician: Schedule an appointment with your child's Paediatrician for an evaluation.

2. Genetic Testing: Discuss the possibility of genetic testing with your doctor to confirm the diagnosis.



Next Steps to Support a Child with Down Syndrome:

Supporting a child with Down Syndrome involves a holistic approach that includes:

Early Intervention: Enrol your child in early intervention programs that focus on speech therapy, physical therapy, and developmental support.

Special Education: Explore educational options, such as inclusive classrooms or specialised programs tailored to your child's needs.

Medical Care: Regular check-ups with healthcare providers to monitor physical health.





Therapies: Occupational Therapy and Speech Therapy can be beneficial.

Support Groups: Join local or online support groups to connect with other parents facing similar challenges.



Strategies for Children with Down Syndrome:

- Provide a structured daily routine.
- Use visual aids and schedules to help with organisation.
- Encourage independence in daily tasks.
- Promote social interactions with peers.
- Focus on speech and communication development.
- Use positive reinforcement for behaviour management.
- Break tasks into smaller, manageable steps.
- Incorporate sensory activities to support sensory needs.
- Encourage physical activity to improve muscle tone and coordination.
- Offer opportunities for creative expression through art and music.
- Use age-appropriate educational materials.



- Emphasise reading and literacy from an early age.
- Teach self-care skills, such as dressing and grooming.
- Foster a loving and supportive home environment.
- Celebrate achievements and milestones.
- Advocate for inclusive educational settings.
- Seek professional guidance when needed.
- Be patient and understanding.
- Focus on strengths and abilities.
- Promote a healthy lifestyle with balanced nutrition and regular exercise.



Remember that every child with Down Syndrome is unique, and their needs may vary. Please reach out to Inspire Allied Health and Education Group to find out how we can support your child and family.



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