Indian Academy of Pediatrics (IAP)

GUIDELINES FOR PARENTS

Care of a Child with Down Syndrome

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9 FAQs on CARE OF A CHILD WITH DOWN SYNDROME

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Care of a Child with Down Syndrome

Q1

What is Down syndrome?

Down syndrome (DS) is a condition, where children with special needs are born with extragenetic material. We all are born with 23 pairs of chromosomes within each cell of our body, numbered from 1 to 22 and X and Y for easy recognition. Normally, every person has a pair of each chromosome. Children with DS have an extra copy of chromosome number 21, that is three in place of two.

All over the world, approximately 1 in 800 children are born with DS. These children differ slightly and need a little more care as compared to normal children to realize their potential. DS is the most common known cause of somewhat reduced intellectual capacity among children.

Every child is different from another child. Due to the difference in their genetic material (chromosomes), children with DS learn to walk and talk later than other children of the same age, but finally achieve these skills. However, their intellectual and physical capabilities have mild-to-moderate delay as compared to other children. They may have difficulty with reading and writing, but they like music, are pleasant, are liked by others, and have a friendly nature.

Q2 How is my child with Down syndrome different from other children?

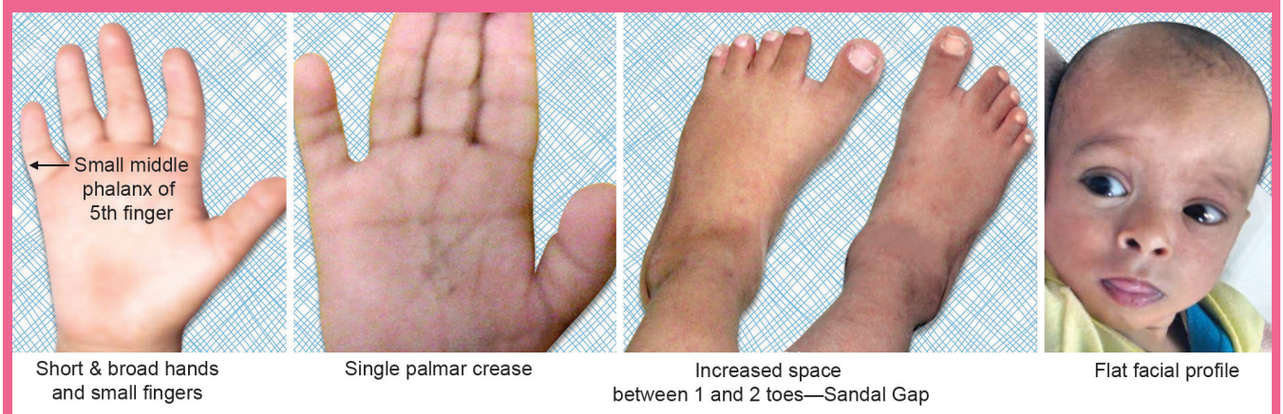


Fig. 1: Pictorial representation of clinical features in children with Down syndrome.

Most children with DS need some special care for their daily needs and schooling. They are slightly shorter than their siblings and parents. Some may look slightly different from family members but look similar to other children with DS. **Figure 1** shows some of the features in children with DS. Children with DS tend to grow slowly but they age faster than usual. They have changes of old age at 40–50 years which other people may have at 60–70 years.

If Down syndrome is a genetic disorder, how did my child get it?

Q3

Down syndrome is usually not inherited from parents and rarely runs in families. Egg cells in mother or sperm cells in father normally carry only one copy of each chromosome. Sometimes when these are formed, one of them receives an extra chromosome 21 from its parent cell when other chromosomes are normal in number. When this egg/sperm gets fertilized by one carrying the usual one chromosome 21 from the other parent, the baby will have three chromosome number 21, causing DS. There are other mechanisms that can cause DS but happen rarely. These changes can occur by chance in any person without any known cause. Mothers more than 35 year of age at delivery have a somewhat higher risk than younger mothers of having a child with DS.

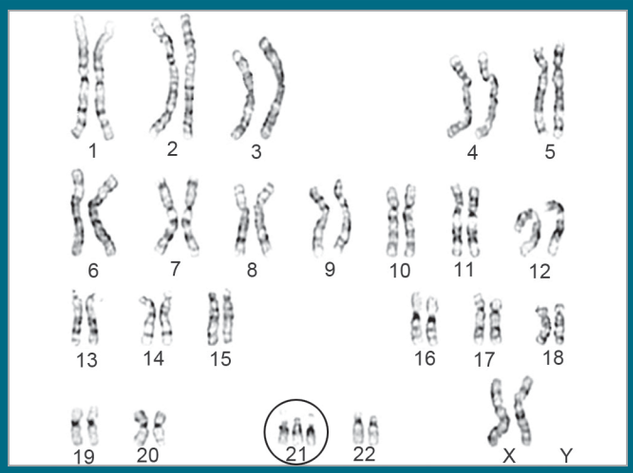


Fig. 2: Karyotype of a female child with Down syndrome (DS) (found in 95% children with DS). Note that all chromosomes are two each in number except chromosome number 21, which are three in number.

However, most children with DS are born to younger mothers as it is their age to have children and many older mothers have children who do not have DS. All pregnant women, irrespective of their age, should be given information about screening for DS.

Down syndrome can be confirmed by a test called "karyotype" (chromosome analysis) that should be done in all children with features of DS to confirm the diagnosis and find the type of chromosomal change causing DS. 95 out of 100 children with DS will have three 21 chromosomes (total 47 chromosomes in place of 46) in the test report (**Fig. 2**). About 1% DS children have two cell lines (mosaic) and 4% have other forms of chromosomal changes (translocation). All children have similar features. For children with DS due to translocation, karyotypes of parents are recommended. Report of karyotype test usually becomes available in 2–3 weeks.

Q4

What are the common medical issues that a child with Down syndrome has?

- Most babies with Down syndrome (DS) do not have good muscle tone. This makes it harder for them to hold neck, learn to roll over, to sit up and to walk but ultimately they are able to do it. Physical therapy can help with these problems.
- Some babies have eye problems such as cataract, watering from eyes, refractory errors, or squint. Simple eye care and use of spectacles will help to combat some of these problems. Surgery may be needed for cataract and squint.

- Children with DS may have cold, ear infections, and sinus infections more often than other children.
- They are more likely to have thyroid problems and hearing issues. Thyroid problems require supplementation with thyroxine. For hearing issues, grommets/hearing aid may be required.
- These babies may also have some kind of heart defect. An *echocardiography* evaluation of baby's heart will show if there is any problem. Surgery can fix the heart problems.

- Some babies with DS are born with blockages in their stomach or intestines (bowels). Some of these may be detected in antenatal scans also. Surgery can fix these problems too.
- Some children have a propensity to develop joint dislocation at neck that needs to be ruled out before participating in any physical training or sports.

Q5

Which health checkups should be done and how frequently?

Children with DS require time-to-time health checkups that are listed in the **Table 1**.

Table 1: Health checkups for children with Down syndrome.

What to check?	*How to check?*	*When to be done?*
Growth	Weight, height, and head circumference*	Every 3 months during first year and then every year
Ear evaluation	BERA/OAE for hearing assessment/ tympanometry	At least twice in first year, then every year
Eye evaluation	Cataract/refractory error	• Every 6 months in first year • Every year till the age of 5 years • Every 2 years till the age of 12 years • Every 3 years >12 years
Thyroid profile	T4 and TSH (thyroid hormones)	• At initial contact • At least at 6 month and at 12 months in first year and thereafter every year
Heart	ECG and ECHO	At initial contact Then as per need
Sleep	Polysomnography (sleep studies)	Sleep studies for all by 4 years
Blood	Complete blood count	After 6 months in first year and then annually till 12 years of age

*Growth charts for Down syndrome.
(BERA: brainstem-evoked response audiometry; ECG: electrocardiogram; ECHO: echocardiogram; OAE: otoacoustic emission; T4: thyroxine; TSH: thyroid-stimulating hormone)

Q6 Is there a cure for Down syndrome (DS)? What do you mean by early intervention program?

There is no medicine to cure DS but therapies are available for a child with DS to help them achieve a productive life at par with others. Early intervention program means to start the therapies *as early as possible*. Sooner you start training the child, better would be the outcome. The capacity to learn new things is maximum till 6 years of age.

- *Occupational therapy (OT)*: To empower the child to do his/her routine independently is OT. Training the child for fine hand movements (**Figs. 3A to D**) such as buttoning, turning page, tearing, holding, folding, tying, etc., are part of OT.
- *Physiotherapy (PT)* (***Fig. 4***): Physiotherapy is very important to achieve neck holding, rolling over, sitting, standing, and walking. It should be started when the muscles are strong enough to bear the body weight. If tried prematurely, it results in premature degeneration of joints, bending of vertebral column, and abnormal gait.
- Persons with DS frequently have flat foot. Using medial arch in sole (**Fig. 5**) is a must while walking.
- *Speech therapy (ST)*: Weak oral muscles, thick and short tongue, narrow oral cavity, and breathing difficulties result in delayed development of speech. ST includes mouth muscles exercises such as blowing (**Figs. 6A and B**), sucking, licking, and chewing.

Figs. 3A to D: Occupational therapy.

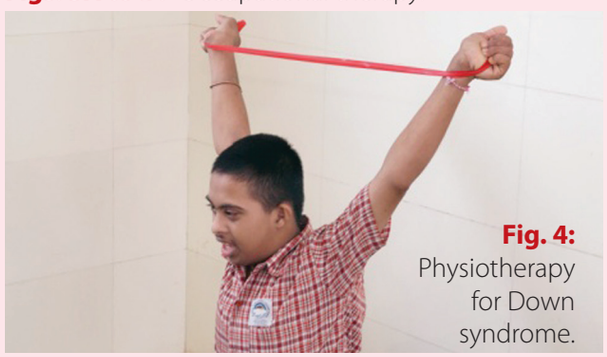


Fig. 4: Physiotherapy for Down syndrome.

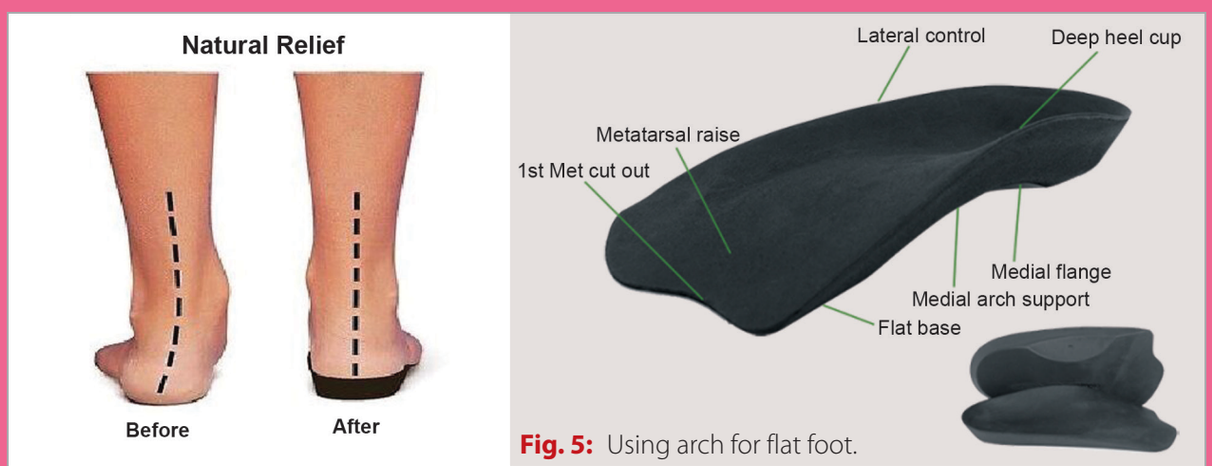
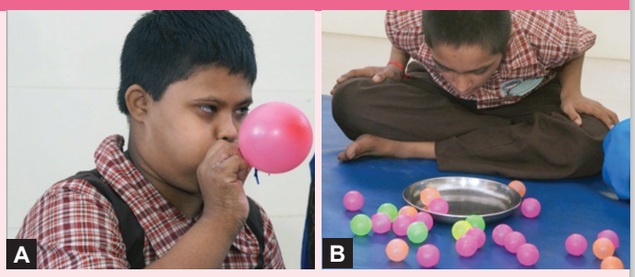


Fig. 5: Using arch for flat foot.

- *Sensory integration (SI)*: We learn through our senses. SI is about stimulating vision, hearing, taste, touch, and smell senses. Exposure to colorful objects, different frequency sounds, tastes, texture, smell, and body movements is helpful in improving these senses.
- It is important to refer these children with Down syndrome to a team involving developmental pediatrician, occupational therapist, physiotherapist for a coordinated and holistic care.



Figs. 6A and B: Mouth muscle exercise.

Q7 How can I help my child with Down syndrome?

- There is nothing to be ashamed of nor have a guilt feeling if one has a child with DS.
- These children are trainable, educable, and can be trusted as employees.
- They are very friendly, likeable, and social.
- They can be trained for self-employment and can lead an independent life with some support.

To achieve best out of them, they need to be nurtured in a supportive home environment involving both parents and the family. Treat your child similar to the other family members. Keep your child connected with his/her environment continuously. Communicate and interact with him/her as much as you can. These are the key factors to speech and language development.

Q8 What is the risk that my next child will have DS?

A family that already has a child with DS or a mother whose age is more than 35 years have higher chances (risk) than other families to have a child with DS. Your risk can be checked from the "karyotype" test done for your child who has DS. Presence of three complete chromosomes 21 indicates a recurrence risk of approximately 1% only. However, if the first child with DS has different type of chromosomes called "translocation", karyotype test will be required for both father and mother. Risk of recurrence is higher, if either of the parents have translocation. If they are not carriers, the risk of recurrence of DS in siblings is 1%.