

Indian Academy of Pediatrics (IAP)



## GUIDELINES FOR PARENTS

# Birth Defects



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### 10 FAQs on BIRTH DEFECTS

1. What is a birth defect?
2. What are the common visible birth defects?
3. How common are the common birth defects?
4. What causes the birth defects?
5. Which children are at risk for birth defects?
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9. What is in future for children with birth defects (prognosis)? And what treatments are available to children with birth defects?
10. How to prevent birth defects in my child?

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# Birth Defects

**Q1****What is a birth defect?**

A birth defect (congenital anomaly) is a health problem or an abnormal physical change that is present when a baby is born.

**Q2****What are the common visible birth defects?**

More than 4,000 different kinds of birth defects are reported which range from minor defects that need no treatment to serious defects that cause disabilities or require medical or surgical treatment. Of these defects in the development of brain or spinal cord, cleft lip and palate and club foot are most common.

Q3

### How common are the common birth defects?

Birth defects constitute the fifth largest cause of death in the newborn babies in our country. Globally, congenital anomalies affect 2–3% of births. In India too, 1–3 out of 100 babies are born with birth defects accounting for 27 million babies being born with birth defects each year.

Q4

### What causes the birth defects?

Birth defects can be caused by certain genes or changes in genes. The genetic defect is inherited from either father or mother or result from sudden changes in genes called as mutations. It can also occur due to maternal exposure to drugs, chemicals, infection or radiation while the mother is pregnant or a combination of factors. However, in majority of cases (70%), the cause of birth defects is not known. Most mothers who give birth to babies with birth defects are healthy without major risk factors and have a normal pregnancy.

Q5

### Which children are at risk of birth defects rather than for birth defects?

Mothers with certain risk factors are at an increased risk of having a baby with a birth defect. These include older age (beyond 35 years), previous baby with birth defect, presence of a family or personal history of birth defects, medical conditions such as poorly controlled diabetes, use of medications around the time, she conceives the baby or in the first few months of becoming pregnant. Mothers who use recreational drugs such as cocaine, marijuana, or drink alcohol during pregnancy are also at higher risk. The risk for certain mutation in genes and thus the risk of birth defects is higher among certain ethnic communities and when parents are related by blood (consanguinity).

Q6

### How are birth defects identified in a child?

After birth, a newborn baby is examined in detailed to look for visible birth defects such as cleft lip, palate, open defects of the spine, defects in arms, legs, and facial features. Some others such as defects of the heart, brain, eye or internal organs, bones, and hearing loss require special tests, such as ultrasound scan, Echo scan, X-rays or hearing tests. Defects of internal organs are suspected based on external features in the baby, clinical examination findings or based on maternal history or risk factors.

Birth defects can also be detected during pregnancy. Screening tests offered in first and second trimesters of pregnancy can indirectly indicate abnormalities or ultrasound scans that can visualize structural defects. Women with risk factors and those with abnormal screening tests are offered confirmatory tests that involve taking a sample of placenta or amniotic fluid for detailed testing. Despite these tests, certain birth defects might not be diagnosed until after the baby is born.

Q7

**What are the symptoms of birth defects in a child and what are the long-term challenges?**

Not all defects will have an impact on the child. Many are only cosmetic issues without any long-term disabilities. Symptoms or problems associated with birth defects will depend on the part of the body affected, extent of involvement, role of the particular part, and association with involvement of other organs. Hence, the challenges could be different in each baby. External deformities are earlier identified during the routine examination, but internal defects might present at various time periods. These are the some of the defects and the challenges posed by them (**Table 1**).

**TABLE 1:** Immediate and long-term impact of part of the body.

<i>Part of the body</i>	<i>Immediate impact</i>	<i>Long-term impact</i>
Head shape	<i>Very large head:</i> Difficulty during delivery	<i>Very small head:</i> Intellectual disability
<i>Lips and mouth:</i> Example: Cleft lip or palate	Difficult in feeding	Difficulty in speech development
Absence of ears or abnormal shape	Cosmetic	Hearing problems
Abnormal fingers, hands, toes, or feet	Cosmetic, challenges during the development of holding or walking	Deformities leading to impaired skills and mobility
<i>Spine:</i> Exposed to exterior	Infections	Difficulty in limb movements
Heart defects	Life-threatening, lack of oxygen leading to bluish color of the skin	Need of corrective surgery
<i>Kidney:</i> Enlarged areas or cysts	Disturbances related to body salts	Poor growth and infections

Q8

### How can I help my child live with a birth defect?

It is common for parents of babies with birth defects to feel stressed and dejected. Every child is unique and has special abilities. So parents should focus on their child's abilities to help them overcome the challenges. The early years are important for the child's brain development and children with disabilities need special attention.

Parents should know about their child's medical issues and the need for continued follow-up and rehabilitation. Communicating with parents having kids with similar condition might help them overcome the stress and offer support to the child. There are parental support groups for Down syndrome and many other conditions. Growing children must receive special attention to aid learning and avoid challenges of bullying in the school.




Q9

### What is in future for children with birth defects (prognosis)? And what treatments are available to children with birth defects?

Treatment of birth defects depend on the severity and the body part involved. Some are minor and easily corrected like the cleft lip and palate while others are major requiring surgery. Some are not amenable to treatment like chromosomal defects or brain defects. Many of these children continue to need specialist care later in life as well as physical therapy, assistive devices or speech therapy. More importantly parents and physicians should help these children to lead a happy and productive lives in the society.

**Table 2** gives information of incidence that how common these defects are and the outcomes of common congenital defects.



**TABLE 2:** Common birth defects.

Congenital defect	Incidence per 100 live births in India (%)	Survival chances till 1 year of age (%)	Intervention	
<b>Central nervous system (CNS):</b> <ul style="list-style-type: none"> <li>Spina bifida</li> <li>Encephalocele</li> </ul>	0.5%	72–92%	<ul style="list-style-type: none"> <li>Surgical repair</li> <li>Multi-team approach</li> <li>Special vocational assistance</li> </ul>	
				
<b>Cardiovascular system (CVS)</b>  <b>Critical congenital heart disease (CHD):</b> Transposition of great arteries, tetralogy of Fallot, etc.	0.5%	55–85%	<ul style="list-style-type: none"> <li>Surgical repair</li> <li>Multi-team approach</li> <li>Special vocational assistance</li> </ul>	
<b>Common CHD:</b> Ventricular septal defect (VSD), atrial septal defect (ASD)	More common in CHD	Normal as general population	<ul style="list-style-type: none"> <li>Medical treatment</li> <li>May need surgical repair</li> </ul>	
<b>Orofacial defects</b>	0.7%			
Cleft lip			92%	<ul style="list-style-type: none"> <li>Surgical repair</li> <li>Multi-team approach</li> <li>Special vocational assistance</li> </ul>
Cleft palate			91%	<ul style="list-style-type: none"> <li>Surgical repair</li> <li>Multi-team approach</li> <li>Special vocational assistance</li> </ul>

Contd...



Contd...

Congenital defect	Incidence per 100 live births in India (%)	Survival chances till 1 year of age (%)	Intervention
Gastrointestinal (GI)	0.4%		
Tracheoesophageal fistula (TOF)		85%	<ul style="list-style-type: none"> <li>• Surgical repair</li> <li>• Multi-team approach</li> <li>• Special vocational assistance</li> </ul>
Anal anomalies		87%	<ul style="list-style-type: none"> <li>• Surgical repair</li> <li>• Multi-team approach</li> <li>• Special vocational assistance</li> </ul>
<b>Musculoskeletal:</b> Congenital diaphragmatic hernia (CDH), omphalocele, etc.	0.7% 	71-90%	<ul style="list-style-type: none"> <li>• Surgical repair</li> <li>• Multi-team approach</li> <li>• Special vocational assistance</li> </ul>
<b>Chromosomal defects:</b> Down syndrome	0.4% 	94%	<ul style="list-style-type: none"> <li>• Occupational assistance</li> <li>• Multi-team approach</li> <li>• Special vocational assistance</li> </ul>

Q10

### How to prevent birth defects in my child?

Incidence of having a baby with some birth defect can be reduced if a pregnant woman takes care of few things. Many of the possible birth defects can be prevented.

Women should take care before conceiving:

- Should take appropriate vaccines regularly.
- Sexually transmitted disease (STDs) should be ruled out before conceiving.
- Before conceiving should start the advised folic acid supplementation.
- Any new or regular medication of postconception should be administered with the doctor's consult.

Obstetrician, pediatrician, and genetic counselor advise is recommended, if the woman belongs to high-risk group. *Example:* If there is a family history of birth defects.

Few of the following precautions are mandatory along with eating healthy and nutritious diet and taking supplements:

- Avoid smoking (active and passive).
- Avoid alcohol.
- Illicit drugs to be stopped or avoided.
- Maintain healthy lifestyle by doing regular exercise and appropriate rest.
- Take regular antenatal check-ups.