

# COMMON BIRTH DEFECTS OPERATED BY DR SANJAY KULSHRESTHA

## [MOST OF THEM OPERATED THROUGH CHARITY UNDER SABD

### BIRTH DEFECTS

**Definition:** Birth defects or congenital malformations are basically structural defects in the body present since birth. However, in broad terms, in addition to the structural abnormalities it also includes any biochemical or functional disorders present since birth.

The structural defects can be external which are obvious on external examination (e.g. hare lip, absent anus, neural tissue swelling on the back, genital deformity etc.) while in other cases the defect could be internal and takes some time to be detected (e.g. malformations of the intestines, kidney, brain, heart etc.).

Some of the birth defects are serious or life threatening needing urgent operation while in others one can wait for some time till the baby becomes fit for surgery.

Total correction of these malformations can be done in one stage in most of the cases. However, in some complex defects the total correction may take two or three stages.

### STATISTICS:

1. About 25-35% of all newborns suffer from birth defects and 3-4% of these are major or serious
2. About 25% of the total deaths all over the world occur during the first year of life and 66% of this mortality occurs in neonatal period (i.e. within one month after birth). This is also a fact that birth defects are one of the leading causes of death among the neonates and account for 25-35% of all neonatal deaths. Thus the need for treating such birth defects can not be over-emphasized.
3. Majority of the birth defects are now completely curable and the baby can look forward to an absolutely normal life.
4. The incidence of serious birth defects is more among the low socio-economic group who cannot afford the highly specialized and expensive neonatal surgery.

With the better understanding and advancements in the pediatric intensive care and anaesthesia, most of the congenital malformations are correctible now. Babies suffering with such problems can now look forward to a normal life and often they do far better than their physicians expect initially.

## COMMON BIRTH DEFECTS AND THEIR MANAGEMENT

Here I am giving a brief introduction of some major congenital anomalies that we commonly encounter in our daily practice. These cases have been divided according to different body systems.

### [I] Pediatric Neurosurgical Problems

1. **Encephalocele** –In this anomaly the membranes or a part of the brain comes outside the head through the defect in the bone. In majority of cases it is situated on the back of the head. Some of them may be large enough to the size of the head itself looking as if the baby is born with two heads [fig-1] while in some it may be small [fig-2]. Those cases having minimal brain contents inside the swelling, having normal headsize and without any other major anomaly in rest of the body, can be treated by operation and carries good results in terms of the functions of the brain [fig-3]. The surgery should be done early as the delay can cause the protrusion of more brain contents outside into the swelling and this brain even if repositioned inside the head may not function properly after the operation.



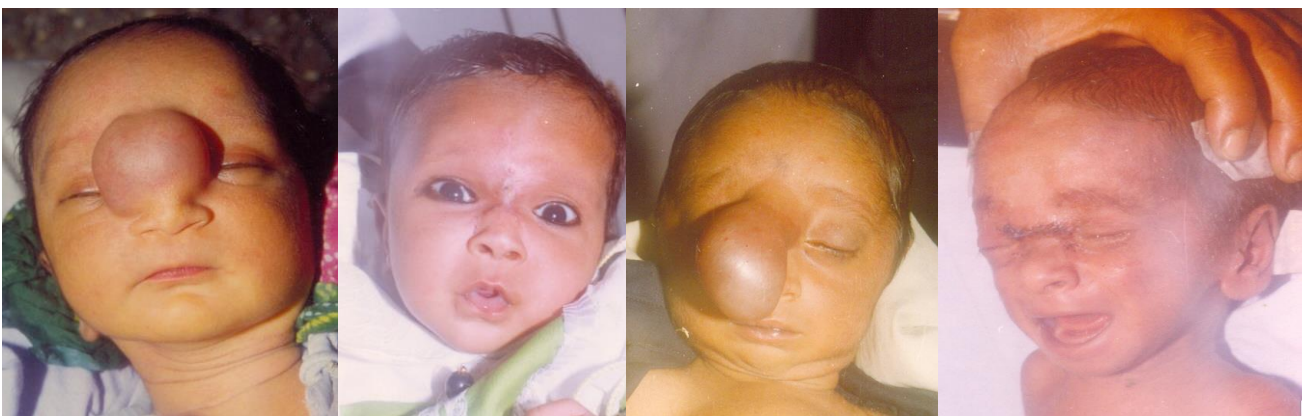
Fig-1



Fig-2



Fig-3↑



Before op

after op

before op

after op



Fig-4 ↑ Showing two cases where the same swelling i.e. encephalocele have emerged from the brain through the front or forehead region. Some of these are large enough [case 2] to resemble the trunk of lord ganesha.

**2. Hydrocephalus:** This is the enlargement of the head due to fluid collection inside the brain [fig-5]. This is either due to excessive formation of this fluid or due to a block in the circulation of this fluid. Results after operation depends on whether there is any other associated anomaly in the brain or in the body and also on the degree of severity of enlargement [mild, moderate or severe]. Most popular operation for this problem is a shunt procedure where an alternate passage is provided for the excess fluid of the brain is diverted into the abdomen. If operated early, majority of cases of mild and moderate categories can lead a normal life.



Fig-5

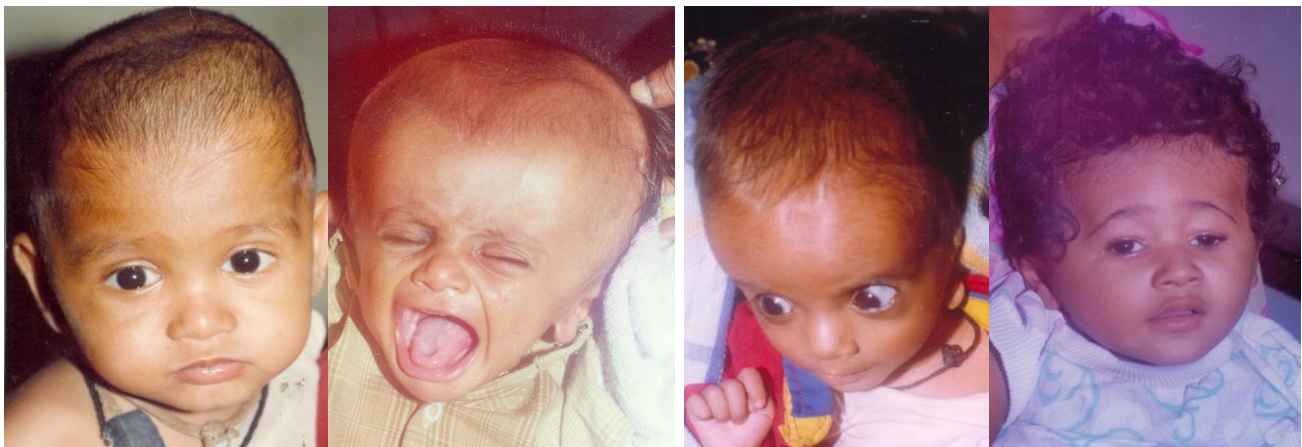


Fig-6 – cases of hydrocephalus after operation

**3. Spina Bifida :** In this condition there is a defect in the backbone or spine through which membranes or nerves or spinal cord can come out. In simple variety the swelling contains only the membranes [fig 7] and it carries good results after operation [fig.8]. There could be more severe variety where the nerves or cord inside the swelling may be damaged causing paralysis in the lower part of the body that makes them unsuitable for the surgery [fig.9]. Earlier correction within few days after birth gives better results.





Fig-7 – simple variety of spina bifida



Fig. 8 – cases after operation



Fig. 9 ↑ Severe variety of variety of spina bifida with paralysis of lower limbs

## **[II] Abdominal Wall Defects**

Some babies are born with a congenital deficiency of the umbilical region so that intestines and other abdominal viscera [e.g. liver] come outside the abdomen. This could be a small defect that can be managed by simple closure [fig-10& 11] while in other the defect may be very large so that almost whole intestines and liver come outside the abdomen [fig. 12]. These large defects are more challenging



to manage. As these all organs comes outside the abdomen during the intrauterine or the in the pregnancy period, the abdominal cavity remains of small size. Due to small capacity of the abdomen, they may require a synthetic mesh [net] to cover the defect [fig. 13] These cases also need an advanced neonatal intensive care as most of them need ventilation or artificial respiratory support after operation. In case if this facility is not available it is better to do such cases in stages.



Fig. 10



Fi



Fig.12 Cases of large abdominal wall defects



Fig. 13 – showing the use of a mesh to cover a large defect [above row] and below are the three such cases after operation.

### **[III] Conjoint Twins**

In this condition the bodies of two babies are joint at some part of their body. This could be a complete twinning where both the babies are more or less fully developed and are joined with each other either through the abdomen, chest, head or the back. Here the target of the surgery is to save both the babies so it requires an extensive preoperative work up and two teams are required to separate them.

The other variety is of partial twinning where one baby is fully mature and the other one is partially developed. The partially developed baby is joined as a parasite to the fully developed baby. I have operated few conjoint twins of incomplete variety that I am showing below.

#### **Case 1 [fig. 14]**

In this case the partially developed baby was joined to the normal baby along the upper half of the body. Both were sharing a common liver. The parasite baby was separated from the normal baby successfully. The parasite baby was found to be having a fully developed lower half of the body [i.e. both lower limbs, genitalia, pelvic and a few abdominal organs].





[Fig. 14]

**Case 2** [fig. 15 A, B & C] showing the photographs of the babies born with a third limb. We also call them as tripus. In these cases the partially developed baby takes the form of a limb [mostly a lower limb]. In cases 15 A & B the third limb was successfully separated from the back bone. In both these cases the limbs were having communication with the spinal cord cavity that required a careful nerve dissection to prevent any paralysis of the lower limbs of the normally developed baby. Fig. 15 C shows two cases of tripus where the parents refused for the operation.



fig. 15 A



fig. 15 B



Fig 15 C

## [IV] Pediatric Tumours and Cancers

Infants and children are not immune to cancers. It can occur as early as in the first or the second month of the life. The cancer in this age group commonly involves kidneys [Wilm's tumour], nerve tissue [neuroblastoma], germ cells [teratoma], liver, muscles, bone, etc. Now with the advancement in the chemotherapy and radiotherapy, the results in such cases have improved significantly. In those cases where the cancer is detected early, the survival rate is more than 80%. A solid lump in the abdomen or in other part of the body should always raise a suspicion of a tumour.

**[A] kidney tumours**] Case 1&2 [ Fig 16 & 17] are the most common solid tumours in children





Fig. 16 pre operative      per operative      post operative [after 4 years]      resected tumour



Fig.17 pre operative      per operative      post operative [after 5 years]      resected tumour

## [B] Tumour of neural tissue [neuroblastoma]



Fig. 18 Pre op      per operative- large tumour is being delivered outside      postoperative



[C] Tumours of germ cells [teratoma] Fig. 19 & 20 These tumors can develop anywhere in the body and can be of very large size. Initially they grow slowly and if not treated timely, they can convert into a frank cancer.

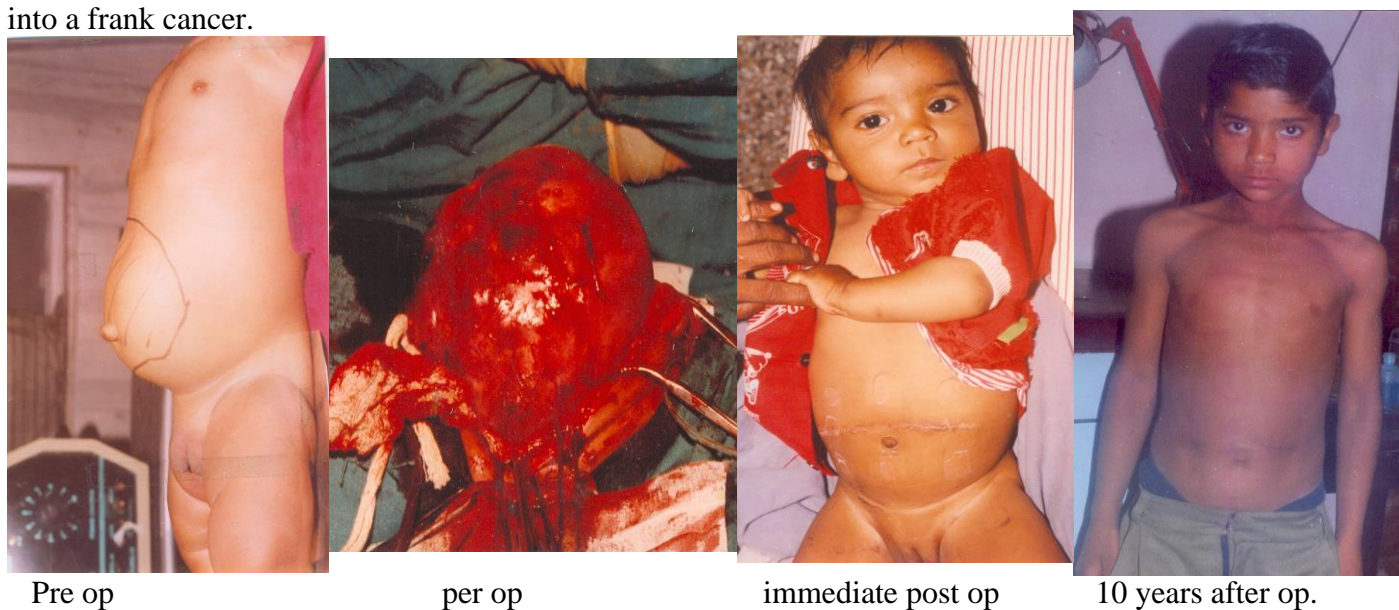


Fig 19. ↑ A case of very large abdominal teratoma operated at the 3 months of age



Fig 20. showing two cases of teratoma of lower back or the sacrococcygeal region. These type of tumours grow during the pregnancy period and present right at the time of birth. In the above case the



weight of the tumour was more than the weight of the baby. Even by caesarian operation it could be delivered with great difficulty. This was excised successfully. In the second case the tumour resembled with that of head as it had full grown hairs and an eye like structure [see arrow] It was also removed successfully. Such tumours turn into a cancerous stage if not treated within 3 months of life.

[D] Tumour of the liver : fig 21. Cancers of the liver are less common in children. Usually they require a major resection. Since the regeneration capacity of the liver is extremely good so even a large resection [upto 85%] is well tolerated by the liver.

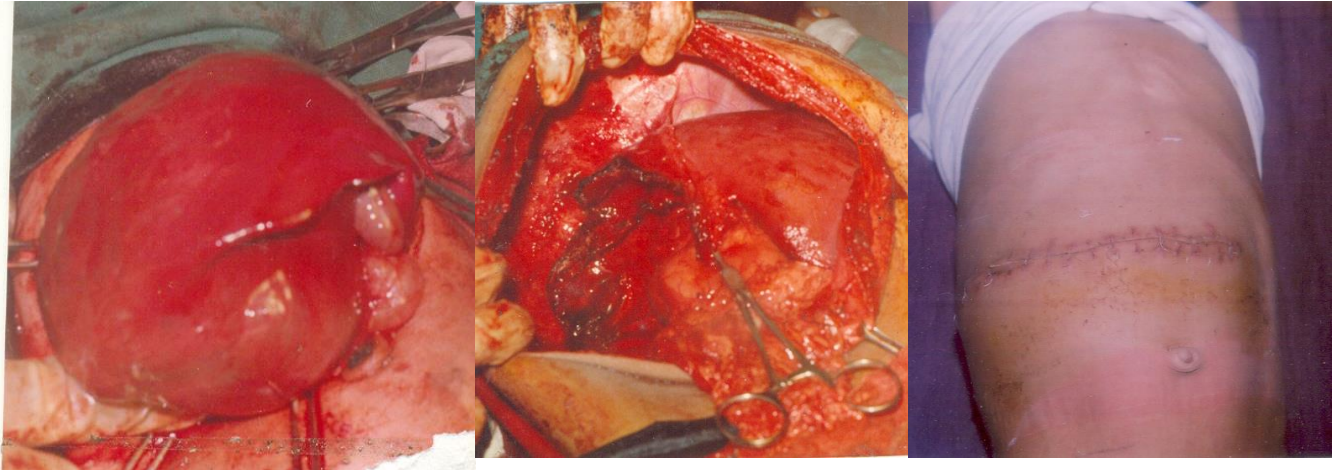


Fig. 21

#### [E] Tumours of the intestines Fig 22

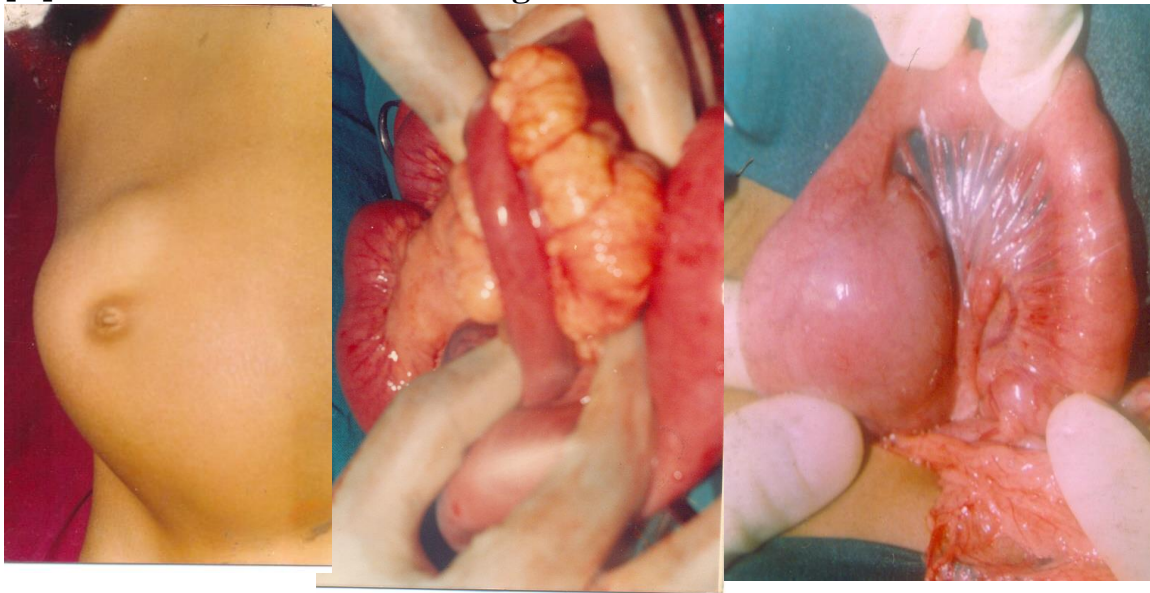


Fig.22

Case of Giant tumour of tongue [fig. 23] - This boy was having a progressively increasing large tumour of the tongue since birth. Due to this he was not able to talk and eat properly and his mouth used to remain open all the time. The parents were extremely poor and could not get their child operated up to the age of 10 years. This tumour was removed successfully and the tongue was reconstructed again. The boy was able to talk and eat properly three months after the operation. He was also able to close the mouth normally.



Fig. 23



## [V] Pediatric Thoracic Surgery

The two most challenging congenital problems of the chest are the [a] absence of the food pipe and [b] diaphragmatic hernia where due to a defect in the diaphragm causes shifting of abdominal organs to the chest cavity. The operation for correction or making the food pipe is the most challenging operation in the neonate. It requires a level III neonatal intensive care. The first successful operation for the absence of the esophagus or the food pipe in the Agra region was performed by me in 1994. Since then more than 150 cases of this problem have been saved. The chances of survival increases if the cases are referred immediately after birth or diagnosed at the time of birth. This is possible only when pediatricians are routinely called by obstetricians to attend every delivery. This anomaly can also be diagnosed during the pregnancy so the arrangements can be made for delivery to be conducted at a better centre where facilities for such operation are available.



## [VI] Genital and inter sex Disorders

There are some congenital defects that involve male and female genital organs so much so that there may be difficulty in deciding the sex of the newly born baby. Whenever a bay is being delivered, the first question that is asked by the relatives is about the sex of the baby whether it is a male or female. If the parents are not able to tell this immediately or changes there version later on, rumours spreads among the relative and in the society that baby belongs to third gender. There are several examples where the babies were forcefully snatched from the parents by the eunuchs. This explains how important it is to determine the sex at the time of birth in our society. So inter-sex disorders are like that of a medical emergency where a quick determination is necessary. This problem is suspected in babies who are born with a very small penis, small vaginal opening, absent testis, open urinary tract, etc. Whether the baby should be is surgically corrected to a male or a female, this decision depends upon the facts: the genetic type of baby, presence or absence of various internal genital organs and what the baby looks externally. The basic fact that is kept in mind before such decision is that during the intercourse female acts as a passive partner while male as an active one. Unless we are able to reconstruct a perfect functioning male organ, we should not opt for rearing the child as a male. Sometimes, if under pressure of parents, we construct an organ that looks like a male penis but does not have an erectile property, can make their sex life miserable. Whether couple would be able to have babies or not, this remains the second priority.



**Epispadias : before and after operation**



