B D R News

The official newsletter of the Birth Defect Registry of India,
(A unit of Fetal Care Research Foundation)

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Proceedings of the First & Second quarterly meetings of the Birth Defects Registry on 06.02.2010 & 08.05.2010 respectively.

The first BDR meeting for the year 2010 took place on 6th February at MediScan Premises, Chennai. Dr.S.Suresh, Director, BDRI gave his welcome address to the Chennai BDR members. He once again spoke on the need to have a uniform protocol for early detection & prevention of anomalies to help reduce birth defects. He mentioned that 11-14 weeks scan is the best modality to diagnose anomalies although it requires a little more expertise than 16-20 weeks scan. It is mandatory to take at least one picture of the cerebellum & cisterna magna while scanning the fetus & compare it with the normal picture. If cerebellum is normal & cisterna magna is visualized, open NTD can be ruled out, he added. With the advent of improved ultrasound facilities, it is ideal to diagnose it at least by II trimester for appropriate follow up action.

While mentioning the present member strength of BDRI, he said that BDR was moving on and Chennai had become richer (with data) with the "Government Institute of Obstetrics & Gynaecology", Egmore, Chennai" contributing data on 18,000 births and the Government Kasthurba Gandhi Hospital, Chennai" on 11,000 births for 2009. 5 new hospitals from Chennai had become members of BDR and had also opted for reporting online. He said that the cream on the cake was the contribution of last 4 months data by Dr. Tripurasundari of Gandhi Hospital, Musheerabad (Hyderabad BDR chapter) with 25,000 deliveries per year. He mentioned his special thanks to Dr. Gio Gnanadurai (Nodal member Sivakasi BDR) with whose help new members from Dhule, M.P and Assam had been enrolled & they would contribute data on 3500 deliveries per year. He also noted that now registry had representation from Bharuch and Rourkela. He had a word of appreciation for Dr. Anu Vij who had added 12 more hospitals to Navi Mumbai Society taking the total number of member hospitals to 52. Dr. Suresh thanked the BDRI team for adding 74,000 more deliveries in the year 2009 taking the number of hospitals to 465 and the number of States covered by BDRI to 18. The welcome address was followed by interesting case presentations made by enthusiastic Chennai BDR members.

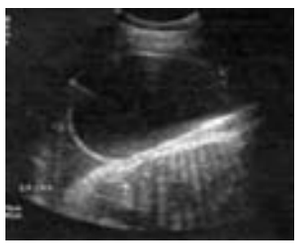
Following are the excepts of the presentations made at the first BDR meeting for 2010

An interesting case of Neural Tube Defect

Dr. M. Mohanambal, Director, Government Kasthurba Gandhi Hospital, Chennai

Dr. Mohanambal presented a case of open NTD diagnosed late at 38 weeks of pregnancy but managed well after birth. She said that the operated child is healthy & attends school at 3 years of age at present.

This child was delivered by a 27 years old mother with an obstetric history of G4P2A2L2. In her current pregnancy, she had her target scan at 20 weeks, when the defect was not diagnosed.



Her first two pregnancies were MTP's by choice. Later at 38 weeks, a large multi loculated cystic mass at C7 & T1 level & dilated ventricles were incidentally picked up.

Since the mass had very little nerve fibre, she was counseled about the possibility of good prognosis following postnatal surgical correction after the baby was born. The baby was naturally delivered & it was found to have a ruptured meningocele in the cervico thoracic region. The baby was immediately transferred carefully with sterile covering in prone position to the surgical ward. Laminectomy & excision of meningocele were done & the wound was closed. Oral feeds were started 24 hours after the surgery was over. The baby was quarantined & was under strict medical vigilance for 3 months

The incidence of NTD was quoted to be 2.7/1000 in the Government hospital. While most of the anencephalic fetuses were diagnosed early & terminated, mothers with bad obstetric history & other risk factors such as diabetes, obesity, epilepsy were screened carefully for neural tube defects. The literature of both open & close NTD were briefly discussed. The lecture ended with a remark that since most women visiting the Government maternity hospitals for antenatal check up are in the third month of pregnancy, awareness about periconceptional folic acid before pregnancy needs to be created among the masses to further reduce the incidence of these defects.

Discussion: A query from the audience was whether the child needed a ventriculo peritoneal shunt? It was explained that the child did not need shunting & the surgical repair was proved to be satisfactory.

A case of congenital ichthyosis

Dr. Anusha- Post Graduate, OBGYN, Government Kasthurba Gandhi Hospital, Chennai.

Dr. Anusha presented a case of Congenital Ichthyosis with literature. The Icthyotic male baby was the second born of a II degree of consanguineous marriage. Family pedigree did not reveal any significant genetic abnormalities in the couple. Their first child was a well girl baby of $1^{1/2}$ years of age.

There were no antenatal scans reported. Mother was admitted with preterm labour at 31 weeks & the baby was born live by assisted breech delivery. He was 1.5 kg & 45cm long at birth. APGARs were low at birth and he was in respiratory distress. He had all the features of a harlequin baby. The skin was dry with edematous & erythematous fissures, scaly, thickened with contractures, distal swelling of the limbs, hyperkeratotic scales on the head, rudimentary pinna, flat nose, ectropion and eclabion. He had a weak cry & died within 3 hours after birth although resuscitation measures were taken. As parents were not willing, skin biopsy and HPE could not be done.

The following is the literature review as reported:

Synonyms: Ichthyosis congenita, keratosis diffusa fetalis & harlequin fetus It was described by Oliver Hart in his diary in 1750 and published in 1896. It was invariably associated with stillbirth or early neonatal death until Lawlor reported a case that survived long in 1985.

Incidence: 1 in 300,000, Mortality / Morbidity: The mortality rate is high. With neonatal intensive care and the advent of retinoid therapy, some babies have survived the newborn period. They are still at risk of succumbing to systemic infection, which is the most common cause of death. **Race:** No racial predilection is known.

Sex: No increased risk based on sex is known.

Genetics: This disorder occurs in consanguineous relationships; multiple siblings within a family can be affected. This has led to the supposition of autosomal recessive inheritance. A new mutation inherited as an autosomal dominant trait has also been suggested.

Clinical features

Skin: Severely thickened skin with large, shiny plates of hyperkeratotic scale is present at birth. Deep erythematous fissures separate the scales.

Eyes: Severe ectropion is present. The free edges of the upper and lower eyelids are everted, leaving the conjunctivae at risk for desiccation and trauma.

Ears: Pinnae may be small and rudimentary or absent.

Lips: Severe traction on the lips causes eclabion and a fixed, open mouth.

Nose: Nasal hypoplasia and erosion of nasal alae may occur.

Extremities: Limbs are encased in the thick hyperkeratosis, resulting in flexion contractures of the arms, the legs, and the digits. Limb motility is poor to absent. Circumferential constriction of a limb can occur, leading to distal swelling or even gangrene. Hypoplasia of the fingers, toes and fingernails has been reported. Polydactyly has been described. Edematous hands clenched in a flexed position is seen.

Genitalia: Small testes and rudimentary scrotum are present.

Temperature dysregulation: Thickened skin prevents normal sweat gland function and heat loss. The infants are heat intolerant and can become hyperthermic.

Respiratory status: Restriction of chest-wall expansion can result in respiratory distress, hypoventilation, and respiratory failure.

Hydration status: Dehydration from excess water loss can cause tachycardia and poor urine output causing acute renal failure.

Central nervous system (CNS): Metabolic abnormalities can cause seizures. CNS depression can be a sign of sepsis or hypoxia. Spontaneous movements may be restricted by hyperkeratosis, making neurologic assessment difficult.

Pathology

All patients with harlequin ichthyosis have ***absent or defective lamellar granules and no intercellular lipid lamellae***. The lipid abnormality is believed to allow excessive transepidermal water loss; lack of released hydrolases prevents desquamation, resulting in a severe retention hyperkeratosis

Types: Based on the immunohistocytochemical features, harlequin ichthyosis has been classified into 3 types.

Treatment: Ensure airway, breathing, and circulation are stable after delivery. Babies require intravenous access. Peripheral access may be difficult. Umbilical cannulation may be necessary. Place infants in a humidified incubator. Monitor temperature, respiratory rate, heart rate and oxygen saturation. Avoid hyperthermia. Once stabilized, transfer newborns with harlequin ichthyosis to a level 3 neonatal nursery. Apply ophthalmic lubricants to protect the conjunctivae. Bathe infants twice daily. Use frequent applications of wet sodium chloride compresses followed by bland lubricants to soften hard skin and to facilitate desquamation. Intravenous fluids are almost always required; neonates initially do not feed well. Consider excess cutaneous water losses in daily fluid requirement calculations. Monitor serum electrolyte levels. A risk of hypernatremic dehydration exists. Maintain a sterile environment to avoid infection.

Retinoids: These agents decrease the cohesiveness of abnormal hyper proliferative keratinocytes. They modulate keratinocyte differentiation.

Rx Isotretinoin: 0.5 mg/kg/d PO.

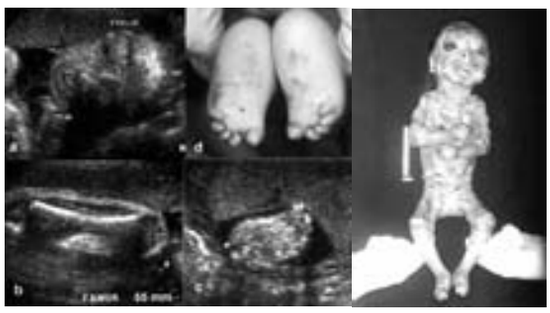
Complications: Gram-positive and gram-negative sepsis has been reported after the newborn period. Children who survive have symptoms that resemble nonbullous congenital ichthyosiform erythroderma, with chronic erythroderma and a fine scale over the whole body. Relapses of severe ichthyosis with eclabion and ectropion occur. Contractures and painful fissuring of the hands and the feet may occur without adequate topical or systemic therapy.

Prognosis: Fulminant sepsis remains the most common cause of death in these infants. Life expectancy is unknown. A report of survival to 24years of age has been published (see below)

Both normal intellect and developmental delay have been described. In general intellectual development is thought to be normal.

Prenatal diagnosis: Fetal skin biopsy can detect harlequin ichthyosis as early as 20 weeks' gestation. Biopsy samples from a number of sites in the fetus reveal the presence of characteristic changes on all skin surfaces, except the mucous membranes. Prenatal ultrasonography can be used to identify

characteristic physical features of harlequin ichthyosis but not until late in the second trimester when enough keratin buildup is present to be sonographically detectable**.**



Amniotic fluid samples obtained as early as 17 weeks' gestation have demonstrated hyperkeratosis and abnormal lipid droplets within the cornified cells. But this is not used in routine practice.

Discussion

Dr. Suresh gave his valuable inputs regarding prenatal diagnosis of harlequin fetuses & other skin anomalies. Fetal skin biopsy is performed around 20 weeks of gestation with a 16 gauge needle & special biopsy forceps. At 20 weeks scan, many of the harlequin features are difficult to visualise & the most commonly used tool is the femur, foot length ratio to diagnose severe type of harlequin & akinesia sequences. Normally they both are same in length at this age & if the ratio is >1(foot is shorter than femur), it is indicative of the anomaly. He recalled a case where the parents refused skin biopsy & the fetus was followed up only with foot length through serial scans. Normal foot length may be indicative of normalcy in the fetus or mild form of icthyosis in high risk cases which may be compatible with life. While taking skin biopsy, representative skin sample can be taken from any part of the body of the fetus. Only in Albinism, sample from a particular site may not reveal the abnormality & hence molecular diagnosis is the best for prenatal diagnosis to rule out this condition. Otherwise EMS (electron microscopic study) of the basement membrane can reveal the characteristic changes in conditions like icthyosis & epidermolysis bullosa. Although molecular diagnosis is available for icthyosis, it is exorbitantly costly but it can help diagnose the problem as early as 11-13 weeks.

Placental chorioangiomas

Dr. Radhika Ramesh, Consultant Fetal Medicine,
Sri.Ramachandra Medical Center, Chennai

Introduction

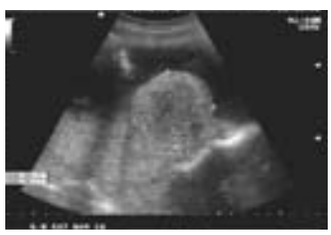
Chorioangiomas are the most common tumor of the placenta, with an estimated prevalence of 1%. They are vascular tumors with the majority being single, small, encapsulated and intraplacental. By virtue of their size, chorioangiomas are very unlikely to be detected during gross pathological examination, unless the placenta is carefully sectioned. Small chorioangiomas tend to remain asymptomatic and do not complicate the course of the pregnancy. Large or giant chorioangiomas, arbitrarily defined as measuring more than 4-5 cm in diameter, have an estimated prevalence varying from one in 5000 to one in 50, 000 pregnancies and are more often diagnosed prenatally by ultrasound imaging.

Complications

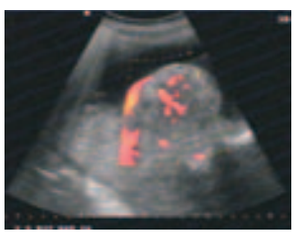
Chorioangiomas have been variably associated with a number of fetal complications including anemia, polyhydramnios, hyperdynamic heart failure, hydrops and growth restriction.

Case study

This was a retrospective observation of all chorioangiomas over a period of 1 year between Oct 2008 to Oct 2009. 4 patients were reported, all with large placental chorioangiomas, more than 5cms in size. 2 of them presented with features of hyperdynamic heart failure and fetal anemia. Since they presented at 31 weeks, decision was made to deliver these babies; they were managed postnatally and they did well. Other 2 patients had no complications with chorioangioma and were serially monitored and delivered at term.



Clinically significant chorioangiomas are larger than 4cms



Well circumscribed placental mass Clearly delineated vascular supply

No	GA at presentation	Size of chorio-angioma	Complications	Outcome
1	31wks (evaluation of polyhdramnios)	6cms	Polyhydramnios, MCA PSV >99th% Cardiomegaly, TR	LSCS live birth 1300gms
2	30weeks (IUGR)	6	Polyhydramnios Cardiomegaly IUGR	LSCS live birth 1150gms
3	34 weeks Diagnosed on routine imaging	5	No complications Weekly monitoring	Induced at 37 weeks 2150gms
4	36 weeks Growth scan done for less fetal movements	5	No complications	Induced at 37 weeks 2500gms

Management options in Chorioangioma

Literature review of a retrospective study over 7 years of 19 cases with giant placental chorioangiomas was presented. This was carried out in a tertiary fetal medicine unit abroad.
The principle reasons for referral were **1)**assessment of polyhydramnios (n=7) **2)**presence of intra - placental mass (n=4) **3)**fetal growth restriction (n=5) & **4)**twin pregnancy (n=3). Associated complications in 18 / 19 cases were variably reported as **1)** polyhydramnios **2)**hyperdynamic heart strain **3)**hydrops & **4)**growth restriction.

Delivery or treatment was considered depending on the severity,type, progression of hydrops as well as the gestational age of onset. Out of 19 cases, 6 did not need any intervention although they were presenting from 23-35 weeks. 3 had polyhydramnios & none had cardiomegaly. Since fetal growth was maintained normally, they were delivered at term. Poly hydramnios could be due to increased urine production, hyperdynamic circulation due to either shunting of blood in chorioangioma or fetal anemia. Fetal anemia could be due to fetomaternal hemorrhage, micro-angiopathic hemolytic anemia, hemodilution & thrombocytopenia. Hyperdynamic circulation & anemia can lead to hydrops.

In another **6** cases, decision for delivery was based on doppler abnormalities, or worsening biophysical profile. When there is fetal growth restriction with chorioangioma, the total functional capacity of the placenta decreases. Large tumor can

act as physiological dead spaces & the residual placenta may be small. If fetal complications occur in late II trimester, delivery may not be the preferred option. Antenatal interventions may be considered. Symptoms like anemia (effect) may be treated or the cause can be treated by blocking vascular supply to the tumor. The rest of **7** had prenatal interventions as: Amniodrainage - 2, Fetoscopic laser ablation - 1, Interstitial laser - 2 (one patient had this twice), Amnio-drainage, transfusion and interstitial laser twice -1 & 1 just had fetal sampling and delivery (GA -31 weeks)

Amnio-drainage was performed if deep vertical pocket was >12cms with tense distended abdomen. However this method offers only a temporary and brief relief of symptoms. This helps us buy time to think of other options.

Fetal blood transfusion was done if MCA PSV was >95th centile. This helped in fetuses suspected to be anemic on MCA-PSV Doppler assessment. In fetuses with high output failure with GA< 32weeks, Fetoscopic laser ablation of surface vessels was done.

When the tumor is close to the cord insertion, **ultrasound guided interstitial laser** may be performed. In the present study ultrasound guided interstitial laser with 17 G needle was used to devascularise the tumor using local anesthesia. It does not require precise placement of needle. There is no usage of toxic substances such as alcohol. This technique can be used even if the placenta is in the anterior uterine wall. With highly vascular tumors, the laser treatment can be repeated after a week rather allowing the risk of excessive thermal damage associated with attempting to stop tumor circulation by one treatment episode. Follow up may be done with 3D power angiogram till the baby is delivered.

Fetoscopic Laser Ablation of the surface vessels may be undertaken if the tumor is in close proximity to the umbilical cord insertion into the placenta to avoid excessive thermal damage to fetal circulation from interstitial laser treatment.

Conclusion

In view of the association between placental chorioangioma and poor pregnancy outcome, close prenatal surveillance should be a routine practice. If complications develop late in pregnancy, delivery should be considered, depending on fetal maturity and the available neonatal support. However, pregnancies with the most severe fetal complications often occur late in the second trimester, at which stage delivery would not be the preferred option owing to fetal prematurity. In these situations, fetal therapeutic options should be considered to optimize the fetal outcome.

Discussion

Case management of a large chorioangioma presenting at 26 weeks with hyperdynamic circulation & persistent fetal anemia was discussed. Since blood transfusions did not help, ablation of the chorioangioma using micro coils was done. But the fetus developed pericardial effusion & had to be delivered at 31+ weeks. The baby could not make it & succumbed in neonatal period. The dictum for correct decision making lies in right case selection & timely intervention. Earlier the presentation worse is the outcome. Prolonged anemia increases cardiac load causing myocardial dysfunction. It was felt that this case would have had a better chance to survive if the procedure had been done 2 weeks earlier when the decision to intervene was being considered.

Diagnostic criteria for ventral wall defects

Dr. Sharada, Consultant Fetal medicine, MediScan

Dr. Sharada explained the importance of diagnosing the relatively common ventral wall defects with the right criteria as their implications vary.

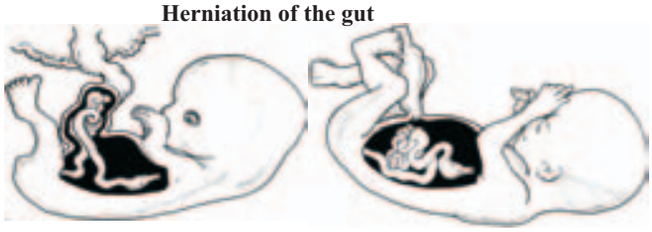
Ventral wall defects are not an uncommon group of anomalies. They constitute 1/2000 live births when detected before fetal viability. Knowledge of a ventral wall defect gives the prospective parents an opportunity to make crucial decisions regarding the pregnancy.

Excellent results can be expected following surgical correction of an isolated ventral wall defect and prenatal diagnosis allows preparation for a planned delivery & corrective procedure with a multidisciplinary approach involving neonatologist, pediatrician and surgeons. On the other hand choice of pregnancy termination can be offered when the defect is a complex one with associated problems.

Embryogenesis

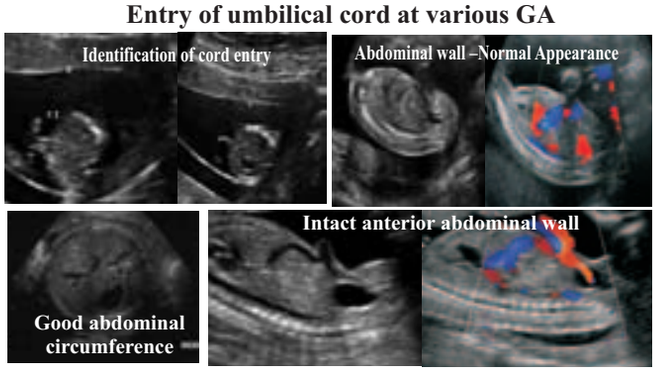
Early in embryonic period, the development of the anterior ventral wall takes place as the embryo folds in both cranial & caudal directions. Physiological herniation of the gut into the base of the umbilical cord occurs between 6-10 weeks of gestation. By 11 weeks the gut takes back its position in the abdominal cavity.

Herniation of the gut



Ventral wall defects can be diagnosed as early as 11-14 weeks by carefully scrutinizing the anterior abdominal wall & seeing the cord entry as seen below

Entry of umbilical cord at various GA

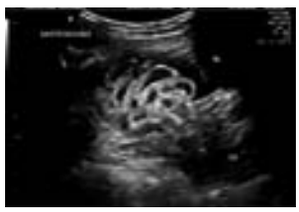


Classification

Ventral wall defects can be classified into 3 groups depending on their spectrum of presentation. They are 1)Gastroschisis 2)Omphalocele with extracorporeal or intracorporeal liver & 3)Midline disruptive sequences comprising of a) Pentalogy of Cantrell b) Ectopia cordis c) Bladder exstrophy d) Cloacal exstrophy & e) Limb Body wall complex. Ventral wall defects are sporadic in nature & hence their recurrence is very low.

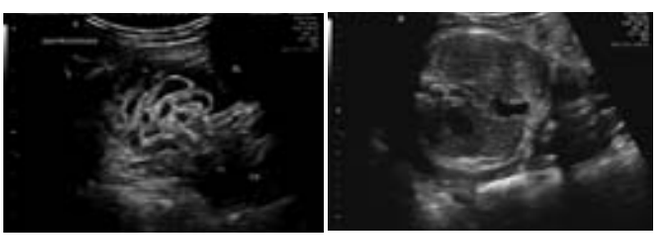
Gatroschisis: In gastroschisis usually the bowel is eviscerated & not the liver as in omphalocele. Since the bowel is exposed to amniotic fluid it appears thickened & in the small bowel there may be obstruction, dilatation or fluid collection. This is a paraumbilical defect seen in the right side of the umbilical cord involving all layers of the abdominal wall. It is usually a small defect in the abdominal wall. Abdominal circumference may not be clearly evident when evisceration is small but the paraumbilical cord entry will denote the anomaly while doing an ultrasound.

One can see eviscerated bowel freely floating in the amniotic fluid with no membrane covering with small bowel, large bowel & stomach. Antenatally it may be visualized as shown.

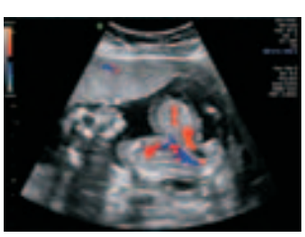


Pitfalls in diagnosis

A few other anomalies like Limb Bodywall Complex, ruptured Omphalocele, Cloacal Exstrophy may mimic Gastroschisis. A case of gastroschisis diagnosed in late III trimester by ultrasonography (USG) turned out to be Amniotic band syndrome (please refer below).



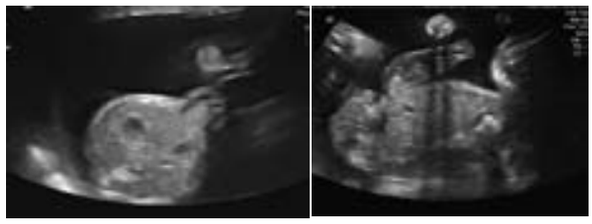
Omphalocele: This is an anterior abdominal wall defect covered with parietal peritoneum / amnion. It is not skin covered. The umbilical cord is seen at the apex of the sac. If the content is extracorporeal liver, intrahepatic umbilical vein coursing the defect may be seen. If the defect is intracorporeal small bowel is usually the content. It can be picked up as early as 12-14 weeks.



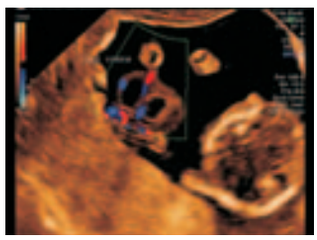
Extracorporeal liver limited by a membrane

Sometimes the membrane will not be clearly seen but the defect will be intact as against the appearance of gastroschisis where the defect will be floating freely in the amniotic fluid. The content of omphalocele may be inferred better by seeing the echogenicity and differentiating between small bowel & the liver as contents.

Pitfalls in diagnosis: Small omphaloceles sometimes may be missed. Physiological omphalocele should not be seen after 12 weeks of gestation. With high resolution transvaginal ultrasound one may pick up umbilical cord cyst which should not be mistaken for an omphalocele. Pseudo defect may be seen in thin patients due to the pressure exercised on the probe while doing an ultrasound. If the fetus has a lax abdomen / poor musculature / narrow thorax as seen in various syndromic conditions, abdominal protrusion may be mistaken for the defect. However demonstration of abdominal circumference & the right entry of the cord will help rule out the problem. In case of umbilical hernia, USG will not be able to pick up the skin or the peritoneal covering, but the herniated bowel will be seen inside the cord & not at the apex or the base of the defect.

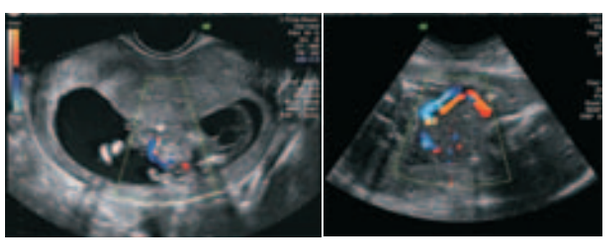


Small omphaloceles can be missed



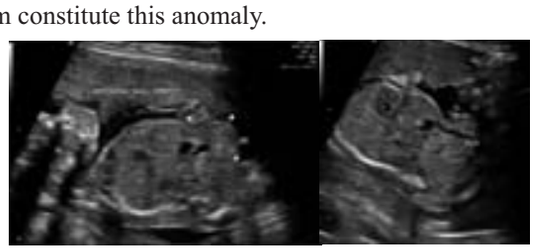
Pentology of Cantrell: This spectrum of midline ventral wall defect comprises of herniation of the heart, bifid sternum, diaphragmatic hernia, pericardial defect & cardiac defect.

Limb Bodywall complex: Although it mimics gastroschisis, it differs from it in many aspects. It is a bizarre anomaly with short or absent cord which is attached close to placenta & associated with cranial, limb defects & scoliosis. There can be single umbilical artery & it is a lethal condition.



Bladder Exstrophy: Incidence of this anomaly has been reported to be low as it has always been overlooked. It can be easily missed on antenatal ultrasound. Non visualization of the normal appearing bladder & normal liquor help us to clinch the diagnosis. Sometimes small cystic structure (urachus) may be mistaken for a bladder. This is generally seen as a soft tissue mass in the lower ventral wall .

Cloacal Exstrophy: Bladder exstrophy with prolapsed ileum constitute this anomaly.



Since there is oligohydramnios, persistent cloaca may be mistaken for cloacal exstrophy.

The lecture ended with a set of 4 questions for self interrogation while diagnosing ventral wall defects antenatally. They are as follow:

1. Is limiting membrane present?
If yes, it may be an omphalocele / umbilical hernia.

2.Relation of the umbilical cord insertion to defect?
If the defect is at the cord insertion it is omphalocele; If paraumbilical it is - gastroschisis; If infraumbilical it may be bladder / cloacal exstrophy; If supraumbilical it is - ectopia cordis.

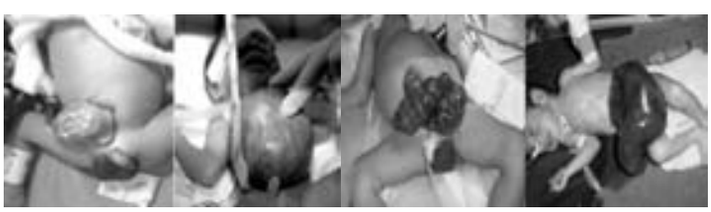
3.What are the organs eviscerated?
If liver is eviscerated, it may be an omphalocele, OEIS complex or Pentalogy of Cantrell. If bowel is eviscerated, it may be - gastroschisis, IC omphalocele / LBWC (limb bodywall complex) or cloacal exstrophy.

4.Is the bowel normal in appearance?
It may present as follows: Gastroschisis -Thickening and bowel dilatation are present. Omphalocele - Bowel is normal in appearance.

Management strategies in abdominal wall defects

Dr. P. Balamourougane, Paediatric Surgeon,
Sri Ramachandra Hospital & RI, Chennai

Abdominal wall defects include Exomphalos, Gastroschisis, Prune Belly syndrome and Exstrophy complex. Dr.Balamourugane discussed the management strategies in the more common ventral wall defects such as exomphalos and gastroschisis. Although exomphalos is more common among the wall defects, the incidence of gastroschisis is found to be on the rise for some unknown reasons. Though they appear similar, the two conditions are quite varied. While the etiology of exomphalos is believed to be failure of lateral folds to fuse, the exact cause of gastroschisis is not clearly established. It may be due to abnormal involution of right umbilical vein, vitello-intestinal arterial accident & ruptured exomphalos are proposed possibilities.



Types of Exomphalos Gastroschisis

The striking difference is the presence of a *sac* (peritoneum, wharton's jelly, amniotic membrane) covering the exposed viscera in examphalos and the *insertion of the cord* on to it, while in gastroschisis, the bowel loops lie free in the amniotic cavity and the cord is inserted onto the intact umbilical ring by the side of the defect. Reactive to the irritant exposure, the loops are covered by a thick *peel*, which contributes to the morbidity.

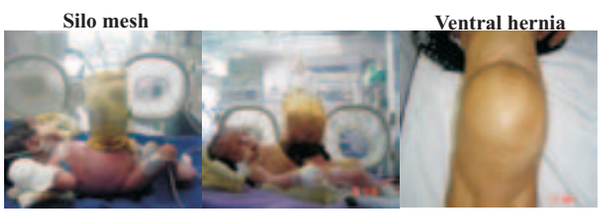
Based on the size and contents, **exomphalos is classified** as Major (>5 cm, Liver as content) or Minor. A small defect with a few loops is called "Hernia of the cord". Gastroschisis is classified as right or left sided, based on the occurrence of the defect in relation to the intact umbilical ring.

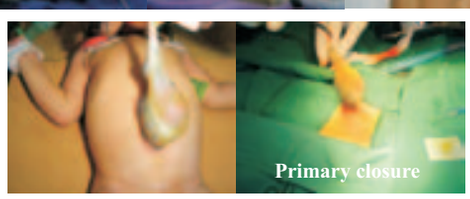
About 40-60% anomalies are **more commonly associated** with exomphalos major, cardiovascular system being the most commonly affected. Chromosomal anomalies and syndromic associations are more commonly seen with exomphalos minor. About 7-30% anomalies are predominantly of the GIT in gastroschisis. However, when the defect is left sided, there is a higher incidence of associated anomalies.

The availability of sophisticated USG has made antenatal detection of the anomalies possible, with well established features. Gastroschisis being an open defect has a higher level of MSAFP elevation than exomphalos. Still it may be difficult in 5% cases to clearly differentiate between the two conditions.

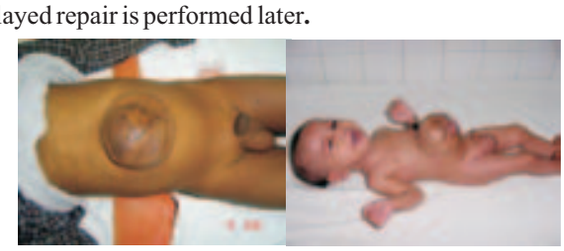
Exomphalos when diagnosed, needs *Karyotyping* to rule out chromosomal anomalies. Once diagnosed, it is safe to shift the baby to a Tertiary care centre for planning delivery and post natal resuscitation and surgery. The route of delivery is dictated by obstetric indication except in exomphalos with the whole liver lying outside, where the risk of injury is high. An earlier delivery in gastroschisis might reduce the severity of the peel. Amnio infusion has also been suggested to improve the same.

The amount of contents lying outside and consequently the size of the abdominal cavity determines the chances of closure. Accordingly a *primary closure, staged closure or a delayed repair* is adopted. Malrotation / lack of normal bowel fixation are universally associated with abdominal wall defects. When the baby is born, the defect is covered with warm saline soaked gauze and a plastic sheet to conserve heat and fluid. Bowel loops are supported and strapped to prevent traction and compression and kept decompressed with NG aspirate and rectal wash outs. Once associated anomalies are ruled out, babies are taken up for surgery. The sac is excised with a longitudinal elliptical incision and the contents reduced with care monitoring the abdominal pressure. Primary closure is done if pressures are acceptable. If it is high even after gentle abdominal wall stretching, the loops are temporarily housed in a *SILO*, created with a mesh and sequentially reduced over 10-14 days. Primary closure is then performed. Sometimes a ventral hernia is created closing only the skin which is repaired later





If the defect is large and occupying the whole abdomen, or in the presence of major anomalies, a conservative approach is adopted. The sac is thickened by the application of escharotic agents - Silver sulfadiazine, mercurochrome, povidone iodine. Delayed repair is performed later**.**



The same principle is adopted in repair of gastroschisis. The availability of spring loaded SILOs has enabled the application of the SILO at bed side in NICU, thus reducing the number of surgical procedures. The management of associated atresia is dictated by the peel. Primary anastomosis may be considered if no significant peel is present, otherwise a delayed repair (to regress the pee l) after a few weeks of TPN (total parenteral

nutrition) support would be the preferred approach. Initial GIT problems and developmental delays are usually made good over a period of 6 months 2 years. The improvement in the intensive support care - both in Ventilation and TPN has resulted in significant improvement in survival rates of these babies, in excess of 90%.

Discussion

Whether size of the omphalocele / gatroschisis determine the prognosis?

Omphalocele when it is small & isolated has good prognosis. Babies do well with just primary closure & they are discharged with a week from the hospital. Gatroschisis in the absence of peel & bowel dilatation when operated do well with good TPN & ventilator support system in the present scenario. Matted peel delays the functioning of the intestines .It was mentioned that there is no need to surgically remove the peel. It would regress on its own within 2 - 4 weeks. Again the commonly reported dysmotility depends on the length of the functioning intestine. If the length is satisfactory with good neonatal support system, the babies are able to catch up growth within 6 months - 2 years of age.

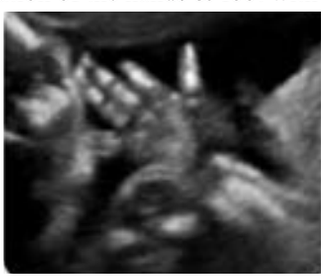
Proceedings of the second meeting of the Birth Defects Registry on 08th May 2010.

Dr. Suresh while welcoming the guests at the second BDR meeting was happy to disclose that the wealth of BDR had gone up by adding 7 more states across the country with its representative members. BDRI statistics year 2010 would reflect sample data from 22 states with 518 member hospitals. He lauded the untiring efforts of the project coordinator in expanding the membership base & he hoped to loop in the neighboring Pondicherry Union & also the far eastern states of India soon. He announced that the present meeting would have the presentation of annual birth defects statistics report for year 2009 & a CME on "Congenital Limb Defects". There was no significant reduction in the incidence of birth defects as per the BDRI data in the last 8 years he added. NTD was still the highest among all. He said that efforts to reduce the most common birth defects by interfacing with the government & educating the professionals at the primary health centers can bring down their incidence by over 60-70%. Chennai members were invited for the Health Expo to be held the following week where FCRF team would try to create public awareness about birth defects prevention. While welcoming the guest speaker Prof. R. Venkataswamy, Plastic Surgeon, Chennai, he said that when a limb defect is antenatally diagnosed, it is wiser to have a Plastic Surgeon along with the geniticist while counseling the family. The excerpts of the presentations are as follow:

Antenatal diagnosis of Hand and Foot anomalies

Dr. Subapriya Kandasamy, Fellow Fetal Medicine, MediScan

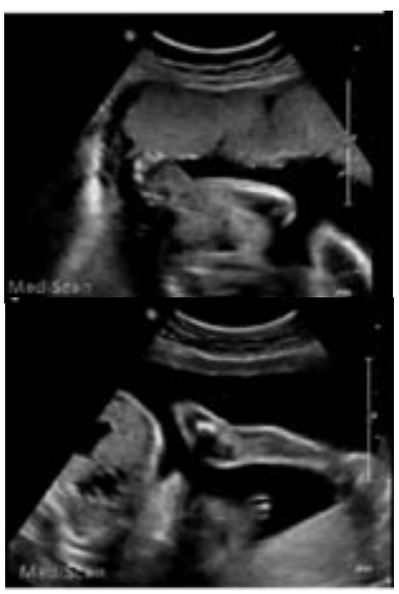
Dr. Subapriya began her lecture saying that congenital limb defects are of functional & cosmetic concern in the affected. Antenatal visualization of the hands & foot with ultrasound was explained. Ossification of humerus, radius, ulna, and phalanges begins at 11 weeks, metacarpals at 12 weeks. The carpal bones ossify after birth. Mild adduction of thumb and atonic fingers are seen in 11th weeks. Appearance of the palm is clear from fingers between 9th-10th week. Fetuses start to clench and unclench their fists from 12th week onwards. Independent movement of each finger is seen from 13 or 14 weeks. **Appearance of hands in various trimesters:** In the first trimester, hands are open and folded in front of the face. In second trimester, hands are held together in front of the face or along the side of the head. In third trimester, they are behind the back or along the side of face and neck.



Diagnosing limb abnormalities

Multicentre RADIUS trial (Smith, 1994) reported a very low detection rate of limb abnormalities (8 - 40%). This may be due to difficulties in fetal position, gestational age, liquor quantity, closed hand, poor penetration and thumb angle. If the angle is different , role of 3D is significant.

The evaluation of fetal limbs, although not currently included in obstetric USG guidelines, is a critical adjunct in characterization of fetal dysmorphology and syndromes, enabling the sonologist to refine differential diagnosis for a particularly affected fetus. Guidelines for screening of limb defects at **18 - 20 week anomaly scan** involve visualization of both forearm bones and the presence of both hands. The detection rate for major limb abnormalities has been estimated at 90%.



Evaluation of limbs for `open' hands and abnormal posturing helps diagnose defects. Open hand helps in diagnosing split hand & transverse defects. Clenched fists rules out syndactyly. Flexion and extension are to be observed in 2nd trimester. Isolated hand and foot abnormalities are difficult to diagnose in 3rd trimester.

Classification of hand abnormalities

Congenital hand deformities may present individually or in combination. The following globally accepted classification is based on their formation, differentiation & their involvement with other systems.

Classification of congenital hand defects

1.Failure of formation of parts:
- a.Transverse arrest
- b.Longitudinal arrest
  - *i.Radial ray defect*
  - *ii.Ulnar ray defect*
  - *iii.Central half defect*
  - *iv.Inter segmental defect (phocomelia)*

2.Failure of differentiation of parts
- ***a.Soft tissue involvement***
  - *i.Disseminated- arthrogyropsis*
  - *ii.Shoulder*
  - *iii.Elbow forearm*
  - *iv.Wrist and hand - cutaneous syndactyly camptodactyly, Thumb in palm deformity, Deviated digits*
- ***b.Skeletal involvement***
  - *i. Shoulder*
  - *ii. Elbow-synostosis*
  - *iii. Forearm - proximal radio ulnar synostosis, distal radioulnar synostosis*
  - *iv. Wrist and hand*
    - *Osseous syndactyly*
    - *Symphalangism*
    - *Clinodactyly*
- ***c. Congenital hormonal condition***

3. Duplication
- *i.Whole limb*
- *ii.Humeral segment*
- *iii.Radial segment*
- *iv.Ulnar segment - Mirror hand*
- *v. Digit*
  - *Polydactyly - Radial, Central, Ulnar*

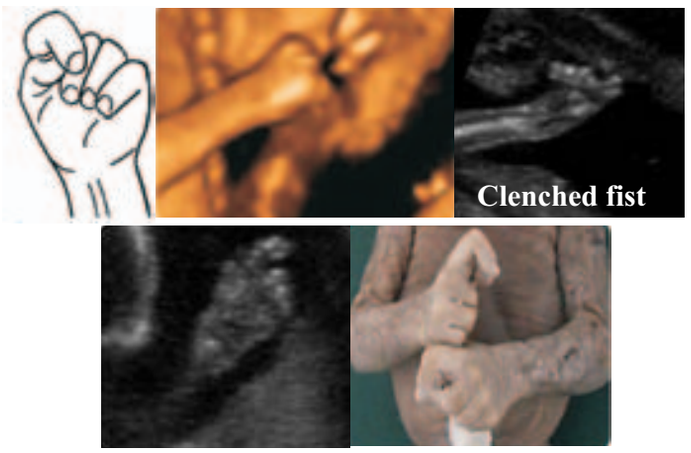
4. Overgrowth
- *i.Whole limb*
- *ii.Partial limb*
- *iii.Digit* - Macrodactyly

5. Undergrowth
- *i.Whole limb*
- *ii.Whole hand*
- *iii.Metacarpal*
- *iv. Digit* - a. Brachysyndactyly, b.Brachydactyly

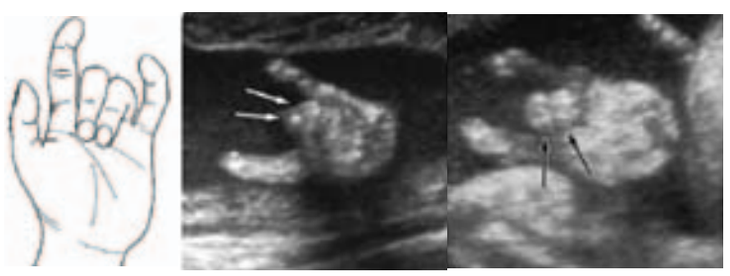
6. Congenital constriction band syndrome

7. Generalised abnormalities

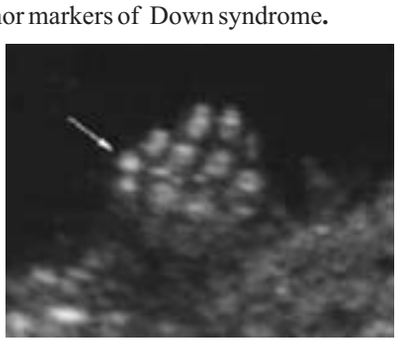
Clenched fist: Index finger overlaps a clenched fist formed by other fingers. PIP (proximal interphalangeal joint) joint of index finger is flexed & ulnar deviated. Thumb is adducted. If this is constant, Trisomy 18 should be ruled out. 30% of trisomies are associated with limb anomalies, the most common being radial aplasia



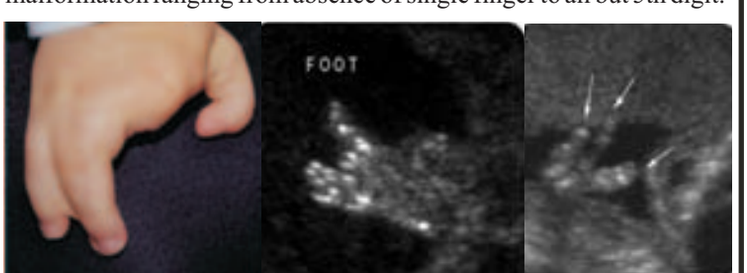
Camptodactyly: Characterised by Flexion contracture of PIP joint of fingers. Severe and multiple digits are suggestive of chromosomal abnormalities (Tri 13,18,15) and multiple contractures. It is often asymmetric, isolated and may progress during infancy.



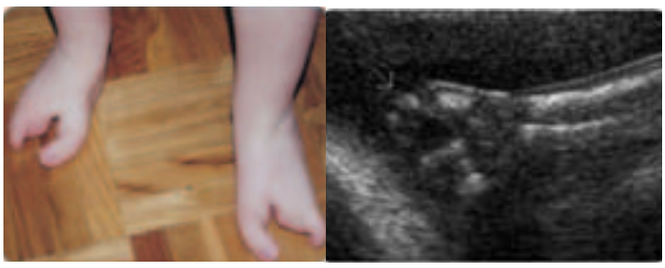
Clinodactyly: Characterised by fixed abnormal deviation of DIP joint of little finger to coronal or radioulnar side due to small middle phalanx of little finger. Familial clinodactyly is isolated and has autosomal dominant trait. About 60% of Down's children have clinodactyly (18% FPR). If diagnosed antenatally one should search for major / minor markers of Down syndrome**.**



Ectrodactyly: Characterised by heterogenous group of hand and foot malformation ranging from absence of single finger to all but 5th digit.



Split hand and foot deformities: This is an autosomal dominant anomaly characterized by the absence of 3rd digit with clefting of proximal hand and foot with syndactyly on either side. There may be absence of both finger and metacarpal bones with a deep V shaped defect. In atypical form it may be a wider cleft formed by defective metacarpals and middle phalanges.



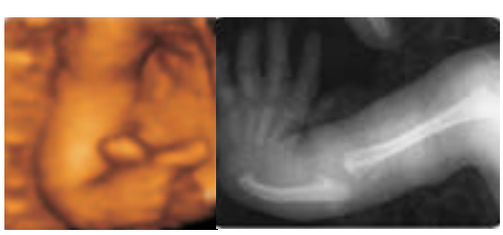
Syndromes associated are split hand, aplasia of tibia, split foot - aplasia of ulna, ectrodactly, ectodermal dysplasia cleft lip/palate syndrome. Early diagnosis of such malformations facilitates better understanding of the defect & bring down the psychological impact on expected parents.

Club hand: There are two types of club hand, they are ulnar and radial club hand.

Ulnar Club hand: It is a rare non syndromic defect secondary to absent / hypoplastic ulnar bone having associations. Diagnosis depends on visualisaiton of radius & ulnar deviation of wrist. It is difficult to differentiate from radial club hand in-utero, as it may be associated with other syndromic anomalies of thumb.

Radial Ray Defects: Radial ray gives rise to first carpal, metacarpal & 2 phalanges of thumb. It is mostly unilateral & sporadic; if bilateral it may be associated with multi system anomalies. Radial ray defects are defined as partial or complete absence of the radius and/or radial ray structures, isolated or associated with other anomalies. If antenatally detected, 3D ultrasound for hand, ear, face, and ECHO are warranted to rule out associations.

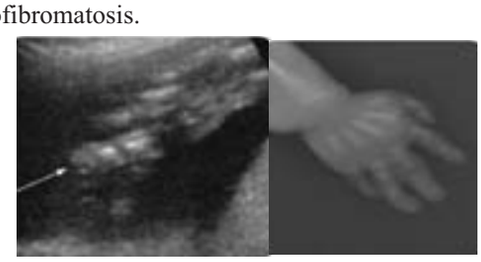
Radial aplasia - index sign



The diagnosis includes thumb hypoplasia or absence, radial carpal hypoplasia or absence. They may be classified as

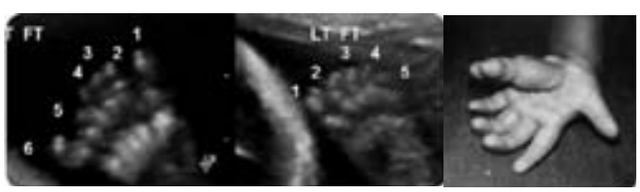
Type I - Short distal radius
Type II - Hypoplastic radius
Type III - Partial absence of radius
Type IV - Absent radius

Macrodactyly: Characterized by overgrowth of all structures in the affected fingers. Radial fingers are more affected. It is usually isolated or associated with anomalies like Proteus syndrome / Type 1 Neurofibromatosis.



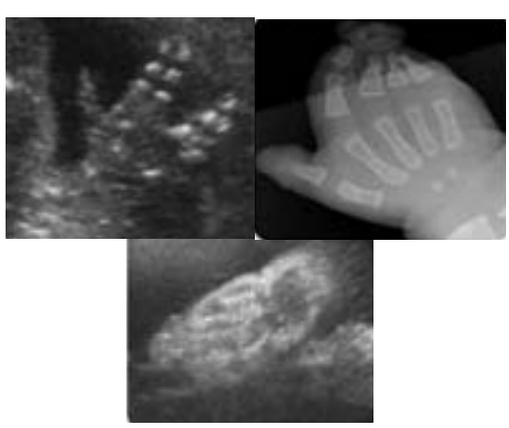
Polydactyly: The fetal finger buds can be imaged by trans vaginal scan as early as 9±10 weeks of gestation & a complete evaluation is possible by 12±13 weeks of pregnancy (Bronshtein et al., 1995). It varies from fleshy to complete digit with flexion and extension or a mere skin tag. Depending on its location it may be called preaxial / postaxial / central polydacytly. It is an autosomal dominant defect if found isolated. Its phenotype is seen in 119 disorders. Isolated fetal **postaxial polydactyly** has favorable outcome. Karyotyping is not warranted for this condition**.**

Preaxial polydactyly is a rare defect & is associated with syndromes of triphalangeal thumb.



Central polydactyly *is also rare & is always an associated defect.*

Syndactyly: Syndactyly is the result of failure of differentiation where fingers fail to separate into individual appendages. The separation of digits usually occurs during 6th & 8th week of embryologic development. It is the most common congenital malformation of limbs. Incidence is 1/2000-3000 live births featuring in more than 28 syndromes. It may be a single gene / sporadic defect or may be caused by teratogens.



Congenital Foot anomalies are of the following types

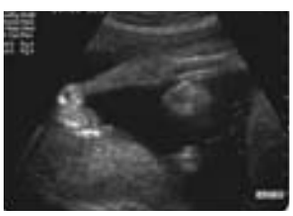
- Club foot
- Polydactyly
- Rocker bottom foot
- Amputation

Clubfoot: Club foot may have following features like adduction of fore foot, inversion of heel, plantar flexion of forefoot, ankle & subluxation of talo-calcaneo-navicular joint resulting in median rotation of dorsal aspect of foot.

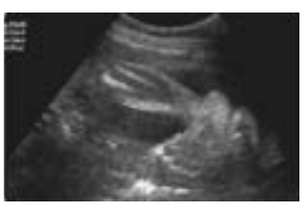
Incidence is 1:1000 live births. Male to female ratio is 2:1.Club foot is an infrequent USG finding and can be both a transient and a late-onset phenomenon. It belongs to a small group of fetal anomalies in which a definitive USG diagnosis may be possible only in the second half of pregnancy. The embryonic development of fetal foot occurs in two phases - Fibular phase (6 - 7 weeks) and Tibial phase (8 - 9 weeks).After 9 weeks, forefoot grows distally and outwards to almost a neutral position. When growth arrest occurs in fibular phase, child is born with a marked club foot deformity.

Depending on the time of diagnosis it may be grouped into 3 types:
1.early onset :12 - 17 weeks
2.late onset : 18 - 24 weeks
3.very late onset: 25 -32 weeks

It has been shown that a fetus can temporarily turn the foot into a position simulating club foot, but this aberrant positioning is usually expected to readjust during a standard examination of 30 minutes. It would appear therefore that 22 to 24week scans should be more reliable for diagnosing club foot, but it may develop even later during pregnancy. Data suggest that about 10% will develop during later stage.

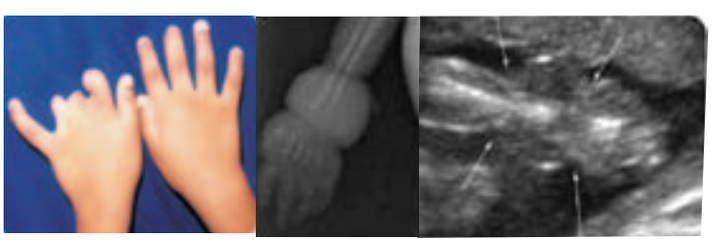


USG finding of metatarsal & phalanges of foot in the same plane as tibia & fibula rules out this problem. Sonographic evaluation of limb movement and muscle mass is also important.



In apparently isolated club foot, it may be the sonographic manifestation of a serious neuromuscular disorder such as arthrogryposis or PenaShokeir syndrome.

Amniotic Band syndrome: Refers to amputation of fingers or limbs to a wide spectrum of associated anomalies. Its prevalence is 7.7/10.000 live births. It is caused by the rupture of the amnion in early pregnancy and "sticky" mesodermic bands from the chorionic side of the amnion causing entrapment of fetal structures leading to decreased blood flow in the constricted limb leading to natural amputation.



Diagnosis: The most common USG finding in amniotic band syndrome is constriction rings of the fingers and toes. Visualization of amniotic bands attaching to a fetus with restriction of motion is diagnostic of the condition.

Management: Depends on the extent of anomalies. Prognosis is good for infants with only minor constriction rings and lymphedema of the digits. Children with amputations of the limbs may require reconstructive or plastic surgery and prosthesis.

Postnatal Surgical management of congenital anomalies of upper limb

Professor R. Venkataswamy, Plastic surgeon,
Apollo FirstMed Hospital, Chennai

Treatment of congenital anomalies has drastically changed in the past few decades such as the technical changes with increasing use of microscopes and magnificent microsurgery, and improved imaging techniques that have permitted earlier and more precise diagnosis of tissue abnormalities. In the future there will be options of fetal correction of abnormalities with refinements in fetal surgery and bio engineering.
Earliest description of congenital anomalies of the upper limb were presented in an illustration as early as 1634 by Chirurgeon Ambrose Parey" (Pare').

Embryology of the upper limb can be viewed in 2 ways: the steps of limb development, and the way the limb is patterned along its 3 spatial axes (proximal to distal, anterior to posterior, and dorsal to ventral axes).

Genetics of upper limb development:
The notochord expresses Sonic hedgehog (Shh), which is thought to regulate the initiation of limb bud formation. The limb bud is an outgrowth of somatic mesoderm (which will form muscles, nerves and vessels) and lateral plate mesoderm (which will form bone cartilage, and tendon) into the overlying ectoderm. This bulging ectoderm called the Apical Ectodermal Ridge (AER) forms at the junction between the ventral and dorsal ectoderm. The time of appearance of the limb bud is at embryonic stage 12, which is equivalent to 26 days after fertilisation and when the embryo is only 4mm long (crown to rump length). Soon after the establishment of the bud, cartilage precursor cells accumulate in the centre (forming the chondrogenesis core) and other connective tissue cells (tendons and muscles) accumulate in the periphery. This cell lineage specification is a function of interacting signals between fibroblast growth factor (FGF) signals and WNT (wingless type mouse mammary tumor virus integration site family).

Clinical Implications of the steps of upper limb development

Complete loss of function of Shh in the notochord leads to severe neurological abnormalities (holoprosencephaly), Cyclopia (single eye), and truncated limbs. This is expected, because the notochord initiates central nervous system and limb development. After the limb bud is initiated, several factors are involved in maintaining it, such as the FGF10, WNT3 / 3a, catenin and p63 expression, Therefore, complete loss of function of Fg10, WNT3/3a, catenin - catenin, or p63 will lead to tetra Amelia. Errors in modulators of the chondrification process result in variable manifestations. Sox - 5/Sox-6 double homozygous mice die in utero because of severe defects in cartilage formation. Mutations in FGF receptors result in acrocephalosyndactyly syndromes (such as Apert and Pfeiffer syndromes). Mutations of noggin (an antagonist of BMP) cause multiple synostosis syndrome, whereas mutations of bone morphogenic protein receptor type IB (BMPR 1 B) cause type A2 brachydactyly (short delta middle phalanges of the index finger and second toe)

Heterozygous mutations of growth and differentiation factor 5 results in brachydactyly type C, whereas homozygous mutations lead to severe disturbances of limb morphogenesis in acromesomelic chondrodysplasisas of the Grebe and Hunter Thompson types.

The classic deformity of Poland syndrome is unilateral aplasia of the sternocostal head of the pectoralis major muscle, ipsilateral short fingers with hypoplastic or aplastic middle phalanges, and simple syndactyly. The exact etiology of Poland syndrome is not known, but it is likely to be a mild ischemic insult during stage 19 of embroyonic life at which time 3 main events occur: 1)development of the sternocostal head of pectoralis major, 2)chonodrificaiton of the middle phalanges and 3) initial separation of the fingers. The steps of **development of the vascular system** have several clinical implications. The earlier development of the ulnar artery might explain the fact that it is usually preserved in congenital ray deficiencies. The radial artery is absent (or severely hypoplastic) in 85 % of cases of radial ray deficiency and in 50 % of cases of ulnar ray deficiency.

The embroyonic insult in transverse deficiencies occurs early (before the development of the ulnar and radial arteries), and hence angiography of the affected limbs shows a persistent median artery.

Limb patterning
A study of limb patterning explains why the hand, for example, has dorsal structures on the dorsum and palmar structures on the palm (dorsoventral axis differentiation); why there is a little finger on the ulnar side and a thumb on the radial side (anteroposterior axis differentiation) and how the limb grows from a proximal to distal direction (proximodistal axis outgrowth). The area specific proteins and transcription factors are controlling these 3 axes.

Each signalling pathway directs limb development along 1 of the 3 axes: proximodistal (from shoulder to digit tip), anteroposterior (from thumb to small finger) or dorsovental (from the dorsum to the palm of the hand).

Transverse deformities: Most common larch is proximal forearm and mid carpal, then metacarpal and humerus, operative indications are very few. Management mainly involves prosthesis filling, status and dynamic orthosis. In mid carpal and metatarsal arrest multiple free toe transfers area option to provide pinch grasp.

Longitudinal inter segmental deficiency (Phocomelia)
Incidence of **phocomelia** increased due to thalidomide ingestion during 1950s. Usual incidence is about 0.8 % of congenital hand abnormalities. Operation indications are few including stabilising proximal joints, lengthening segments of long bones that are present or bone transfer.

Radial club hand: Treatment is stabilisation of wrist by centralisation or radialisation of ulnar &distraction to lengthen the available radius bone. Usually procedure is done at 6 months of age. This would later require pollicisation for thumb ray deficiencies.

Hypoplastic and absent digits: The treatment options are 1) prosthetic replacement, deepening of 1st web digital reconstruction or 2) digital lengthening by microvascular toe transfer.

Cleft hand: It is functionally an excellent hand hence majority of the surgical corrections are done to improve the aesthetics.

Syndactyly: When syndactyly involves only skin and finger tip components, it is considered as **simple syndactyly.** When digits have remained together because of underlying conjoined structural elements or have secondarily joined as a result of amniotic "banding" or are part of syndromes, then it is considered to be **complex syndactyly.** Most frequently the web space involved is the long and ring interspace on the hand -- the second third finger interspace in the hand and the second third toe interspace in foot. Early surgical release is recommended before 4 to 6 months of age especially when it is complex syndactyly or syndactyly involving unequal fingers.

Constriction ring syndrome: Three types of problems are common here amputation, constriction rings and syndactyly. For constriction rings ,early band excision and release combined with Z palsy is the indication. Syndactyly release to be started before 4-6 months of age. **Polydactyly:** Surgical correction is to create a stable optimal composite digit.

Thumb Hypoplasia: This may be isolated or associated with other anomalies especially radial club hand. The management is to do pollicisation of the index finger or Second toe transfer.

Conclusion: Surgical intervention is needed as early as possible otherwise false pattern gets imprinted. The current trend is to operate on them anytime after the sixth months. If warranted even earlier. The surgical treatment nowadays is done as early as 6 months of age. Future depends on prevention of early surgery in the intrauterine stage. A coordinated national registry is essential to document the incidence of limb defects. The speaker ended his lecture with a note of appreciation that the efforts of BDRI to estimate the baseline prevalence of all birth defects under the leadership of Dr. Suresh is laudable.

Annual Birth Defects Statistics Report of BDRI

Ms.V.Jayanthi, Statistician, MediScan

Birth Defects Registry of India Annual report 2009

Introduction

Worldwide the prevalence of congenital malformation is about 2 -3%. In India, though nation wide prevalence estimate is not known, a few small hospital-based studies indicate it would be high. However, there is no systematic surveillance exist for birth defects in India.Having understood the lacunae in this area, Fetal Care Research Foundation (FCRF) established the Birth Defects Registry of India (BDRI) in 2001.

This is the ninth successive annual statistical report of BDRI. It presents the birth prevalence of birth defects estimated from 46 regional registries; viz. Chennai, Erode, Trichy, Lalgudi, Madurai, Nagercoil, Ramanathapuram, Dindigul, Tanjore, Tirunelveli, Vellore and Sivakasi in TamilNadu, Hyderabad, Musheerabad and Nalgonda in Andhra Pradesh, Bangalore, Mysore, Manipal, Dharwad and Belgaum in Karnataka, Trissur and Calicut in Kerala, Mumbai, Pune, Akola, Wardha, Solapur, Latur, Pandharpur, Satara, Dhule, Jalna, Sewagram

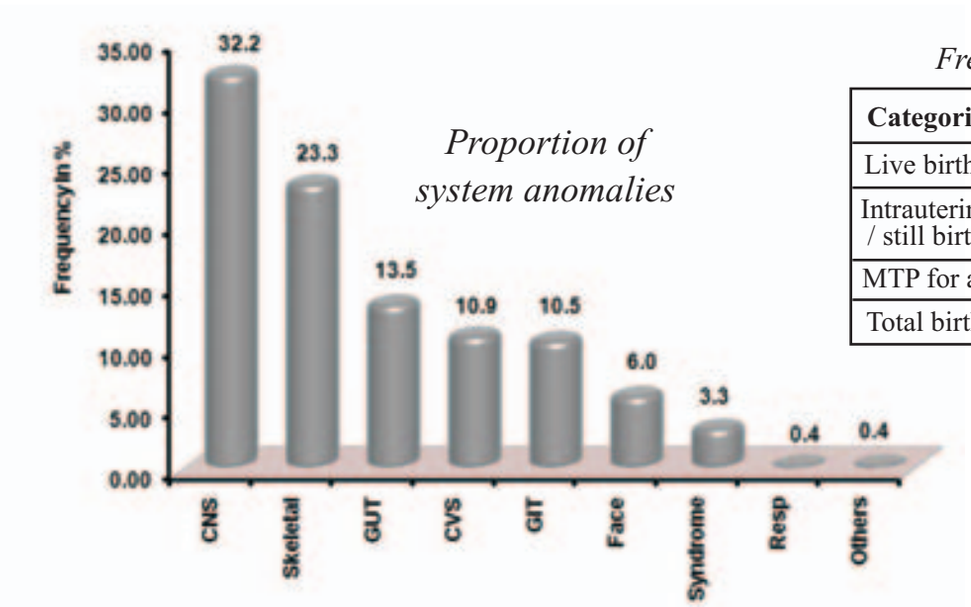
and Jalgaon in Maharashtra and Ahmedabad, Himant nagar, Bharucch and Vis Nagar of Mehsana district, Rajkot in Gujarat, Amristar in Punjab, Varanasi, Agra in Uttar Pradesh, Bhilai in Chattisgarh, Delhi and Goa. Data from Trichy and Lalgudi, Hyderabad, Musheerabad and Nalgonda, Solapur, Latur and Pandharpur were combined because they represent the same geographic area (Administrative district).

Programme description: BDRI is a hospital-based descriptive surveillance program, which passively collects (voluntary reporting) data on structural and chromosomal birth defects from hospitals of defined geographic areas. Collected data are checked by the Dysmorphologist before storing into the database. The diagnostic terms are then coded according to ICD10 version. Finally statistical analysis is done and the yearly annual report is presented to the members.

Results: During 2009 there were 1, 85,849 births reported from the member registries. Of which 97.1% were live born. There were 1750 cases with birth defect (s). The over all crude birth prevalence is 94.2 / 10,000.

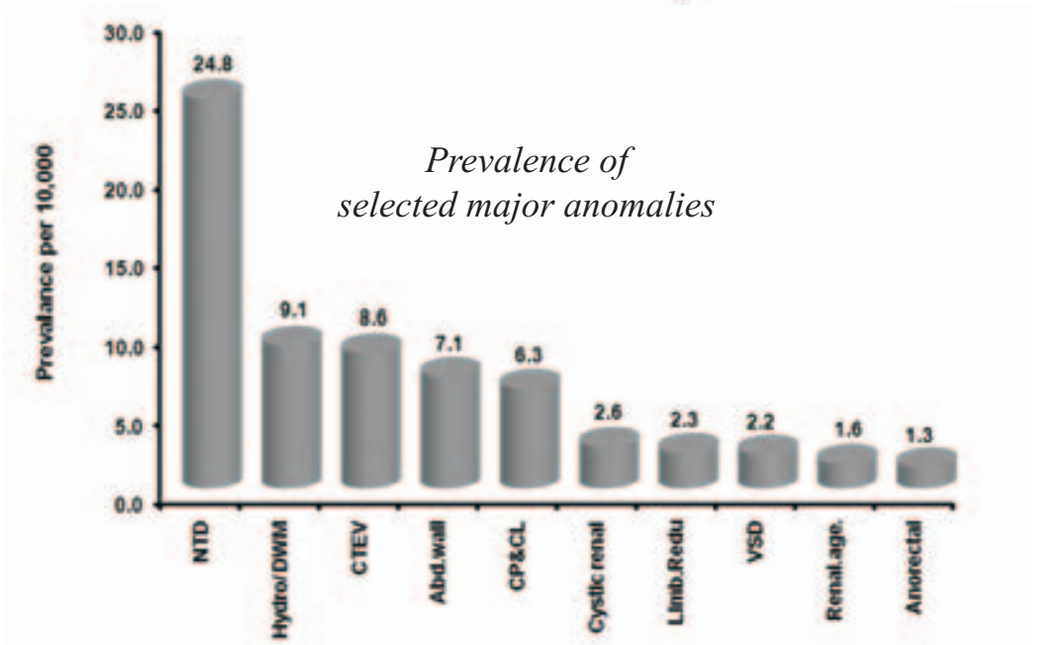
BDRI Statistics -year 2009 Highlights

- The prevalence of Central nervous system & Musculoskeletal anomalies is high when compared to other system anomalies in almost all regions.
- NTD tops the list followed by Hydrocephalus / Dandiwalker malformation & Congenital talipes equino varus while counting the prevalence of the individual system anomalies.

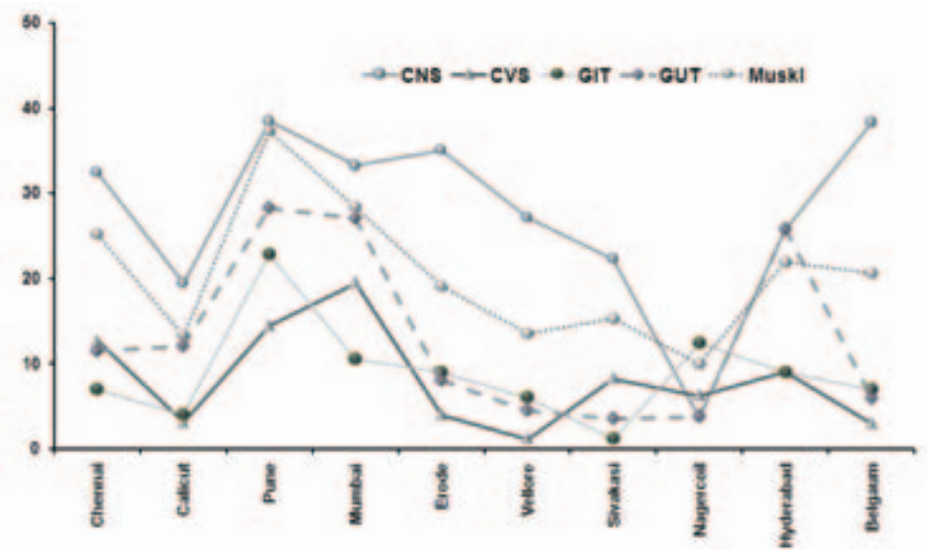


Frequency of birth categories

Categories	N	%
Live birth	180379	97.1
Intrauterine fetal death / still birth	5019	2.7
MTP for anomaly	451	0.2
Total births	185849	100



Crude birth prevalence of selected system anomalies across selected registries



Limitations of the data:

Though the overall crude birth prevalence is 94.2 per 10,000, it is still underestimated since the expected prevalence is at least 2%. The plausible reasons could be, a. Data not population based, (e.g. only 37.3% of total births are covered by the program in Chennai) b. Data collection is not active, c. Most of the minor anomalies might not have been reported, d. Despite using multiple sources of data ascertainment , only a few cases from neonatologists and pediatricians were reported. Though the estimated prevalence may not reflect the true population prevalence in those regions, it shows the pattern and type of congenital malformations most likely to occur in those areas.

BDRI appreciates the following registries of Bangalore Belgaum, Chennai, Calicut, Dharwad, Dindigal, Erode, Goa, Jalgaon, Mysore, Nagercoil, Navimumbai, Pune, Ramanathapuram, Sivakasi, Solapur,Thane, Trissur, Trichy, Visnagar for their data consistency and regularity in sending data to the central registry since their induction.

Table 2A-J: Anomaly specific and system specific crude birth prevalence of congenital malformations across all registries

Diagnostic Grouping	Number of cases	Prevalence / 10,000
A. Congenital Anomalies of the Central Nervous System (Q00 – Q07)	**692**	**37.2**
A01 Anencephaly (Q00.0) **(Incl. Acrania, Exencephaly, Iniencephaly)**	213	11.5
A02 Encephalocele (Q01.0 – Q01.9) **(Incl. Frontal & Occipital Encephalocele/ Meningocele)**	48	2.6
A03 Microcephaly (Q02)	12	0.6
A04 Congenital Hydrocephalus without Spina bifida (Q03.0 – Q03.9) **(Incl. Dandy – Walker malformation, Ventriculomegaly)**	169	9.1
A05 Spina bifida without anencephaly (Q05.0 – Q05.9) **(Incl. Meningocele, Meningomyelocele, Myelocele, Rachischisis, excluding Spina bifida occulta)**	200	10.8
A06 Holoprosencephaly (Q04.2)	30	1.6
A07 All other congenital malformations of brain, spinal cord & nervous system (Q04, Q06 & 07) (Incl. Agenesis of corpus callosum, absence of nerves, cerebral cysts and cerebellar malformations, etc.)	20	1.1

Diagnostic Grouping	Number of cases	Prevalence / 10,000
B. Congenital Anomalies of Eye, Ear, Face & Neck (Q10 – Q18)	**127**	**6.8**
B01 Anophthalmos / Microphthalmos / Macrophthalmos (Q11.0 – Q11.9)	17	0.9
B02 Absent external auditory meatus (Q16.1)	29	1.6
B03 Low set ears (Q17.4)	20	1.1
B04 Anomalies of facial bones	47	2.5
B05 All other congenital anomalies of Eye, Ear, Face & Neck (Q10 – Q18)	14	0.8

Diagnostic Grouping	Number of cases	Prevalence / 10,000
C. Congenital Anomalies of the Circulatory System (Q20 – Q28)	**235**	**12.6**
C01 Common Truncus / Persistent Truncus arteriosus (Q20.0)	2	0.1
C02 Double outlet right ventricle (Q20.1)	6	0.3
C03 Transposed Great vessels (Q20.3)	13	0.7
C04 Ventricular Septal Defect (Q21.0)	42	2.3
C05 Atrial Septal Defect / Patent or persistent foramen ovale (Q21.1)	24	1.3
C06 Atrioventricular septal defect / Endocardial Cushion Defect / Ostium primum (Q21.2)	7	0.4
C07 Tetrology of Fallot (Q21.3)	8	0.4
C08 Tricuspid regurgitation (Q22.2)	5	0.3
C09 Ebstein's anomaly (Q22.5)	0	0
C10 Hypoplastic right heart syndrome (Q22.6)	5	0.3
C11 Other tricuspid valve abnormalities (Q22.8)	0	0
C12 Bicuspid aortic valve (Q23.1)	0	0
C13 Hypoplastic left or Right heart syndrome (Q23.4, 23.5)	20	1.1

C14 Dextrocardia (Q24.0)	3	0.2
C15 Patent ductus arteriosus (Q25.0)	15	0.8
C16 Anomalies of arch of Aorta (Q25.1 & 25.4)	11	0.6
C17 Anomalies of pulmonary artery (Q25.5 – 25.7)	16	0.9
C18 Total anomlaous pulmonary venous connection (Q26.2)	3	0.2
C19 Single umbilical artery (Q27.0)	45	2.4
C20 Other specified and unspecified congenital heart anomalies (Q20.2, Q20.4, Q20.8, Q22.3, Q22.4, Q23.0, Q23.2, Q24.8, Q24.9 & Q25.8, Q26.2, 26.9)	64	3.4

Diagnostic Grouping	Number of cases	Prevalence / 10,000
D. Congenital anomalies of the Respiratory system (Q30 – Q34)	**8**	**0.4**
D01 Congenital cystic adenomatoid malformation of lung (Q30.0)	1	0.1
D02 Absence / Malformation of nose (Q30.1 – Q30.9)	0	0
D03 Laryngeal atresia (Q31.8)	0	0
D04 Tracheal atresia (Q32.1 – 32.9)	0	0
D05 Agenesis of lung (Q33.6, Q33.8)	5	0.3
D06 Other Respiratory anomalies	2	0.1

Diagnostic Grouping	Number of cases	Prevalence / 10,000
E. Congenital anomalies of the Gastrointestinal tract (Q35 – Q45)	**225**	**12.1**
E01 Cleft palate (Q35.0 – Q35.9)	29	1.6
E02 Cleft lip (Q36.0 – Q36.9)	28	1.5
E03 Cleft palate & cleft lip (Q37.0 – Q37.9)	62	3.3
E04 High arched palate (Q38.5)	3	0.2
E05 Other congenital malformations of tongue and mouth (Q38.2, Q38.3)	0	0
E06 Atresia of esophagus without fistula (Q39.0)	18	1.0
E07 Tracheoesophageal fistula with atresia (Q39.1)	0	0
E08 Tracheoesophageal fistula without atresia (Q39.2)	28	1.5
E09 Gastric outlet obstruction (Q40.0)	0	0
E10 Tubular Stomach (Q40.2)	0	0
E11 Absence, atresia and stenosis of small intestine (Q41.0 – Q41.9)	18	1.0
E12 Imperforate anus (Q42.3)	24	1.3
E13 Other Congenital malformations of large intestines (Q42.1)	0	0
E14 Meckel's diverticulam (Q43.0)	0	0
E15 Anomalies of liver and gall bladder (Q44.0 – Q44.9)	8	0.4
E16 Abscent pancreas (Q45.0)	0	0
E17 Anovestibular fistula / Rectovestibular fistula (Q64.7)	0	0
E18 Other specified and unspecified gastriointestinal tract (Q43.1 – 43.9)	16	0.9

Diagnostic Grouping	Number of cases	Prevalence / 10,000
F. Congenital Anomalies of the Genital and Urinary Systems (Q50 – Q64)	**289**	**15.6**
F01 Congenital malformation female genital organs (Q50.0 – Q52.9)	2	0.1
F02 Undescended testis (Q53.0 – Q53.9)	21	1.1
F03 Hypospadias (Q54.0 – Q54.9)	43	2.3
F04 Other congenital malformations of male genital organs (Q55.0 – Q55.9)	22	1.2
F05 Indeterminate sex (Q56.4)	38	2.0
F06 Renal agenesis (Q60.0 – Q60.6)	29	1.6
F07 Cystic kidney disease (Q61.0 – Q61.9) (Incl. Infantile or Adult polycystic kidney and Multicystic dysplasia)	56	3.0
F08 Congenital hydronephrosis (Q62.0)	70	3.8
F09 Pelviureteric junction obstruction (Q62.1)	17	0.9
F10 Other ureter anomaly (Q62.4 – Q62.9)	3	0.2
F11 Other congenital malformations of kidney (Q63.0 - Q63.9) (Incl. Fused / Horseshoe kidney)	8	0.4
F12 Ectopia vesicae / Bladder exstrophy (Q64.1)	0	0
F13 Congenital posterior urethral valve (Q64.2)	4	0.2
F14 Other congenital malformations of bladder & urethra (Q64.3, Q64.8)	10	0.5

Diagnostic Grouping	Number of cases	Prevalence / 10,000
G. Congenital Anomalies of the Musculoskeletal System (Q65 – Q79)	**500**	**26.9**
G01 Congenital dislocation of hip (Q65.0, Q65.1)	8	0.4
G02 Talipes equinovarus (Q66.0)	170	9.1
G03 Other Congenital malformations of feet (Q66.1- Q66.9) (Incl. Rocker bottom foot)	6	0.3
G04 Congenital Musculoskeletal deformities of head, face, spine & chest (Q67.0 – Q67.9) Incl. Dysmorphic face (Q67.0)	2	0.1
G05 Congenital deformities of knee (Q68.2) Genu recurvatum	7	0.4
G06 Polydactyly (Q69.0 – Q69.9)	62	3.3
G07 Syndactyly and polysyndactyly (Q70.0 – Q70.9)	20	1.1
G08 Upper limbs - reduction defects / shortening (Q71.0 – Q71.9)	19	1.0
G09 Lower limbs - reduction defects / shortening (Q72.0- Q72.9)	20	1.1
G10 Unspecified limbs - reduction defects / shortening (Q73.0 – Q73.8)	21	1.1
G11 Arthrogryposis (Q74.3)	5	0.3
G12 Other congenital malformations of limbs (Q74.8 & Q74.9)	11	0.6
G13 Hypertelorism (Q75.2)	3	0.2
G14 Other congenital malformations of skull & face bones (Q75.0– 75.9)	7	0.4
G15 Spina bifida occulta (Q76.9)	1	0.1

G16 Other congenital malformations of bony thorax and spine (Q76.0 – Q76.8) (Incl. Scoliosis, Hemivertebre etc)	24	1.3
G17 Osteochondrodysplasia with defects of growth of tubular bones & spine (Q77.0 – Q77.9)	42	2.3
G18 Osteogenesis imperfecta (Q78.0)	3	0.2
G19 Diaphragmatic Hernia (Q79.0)	49	2.6
G20 Absence / Eventration of diaphragm (Q79.1)	0	0
G21 Exomphalos / Omphalocele (Q79.2)	52	2.8
G22 Gastroschisis (Q79.3)	22	1.2
G23 Thanatophoric Dysplasia (Q77.1)	2	0.1
G24 Other congenital malformations of abdominal wall (Q79.5, Q79.6, Q79.8) (Incl. Limb body wall complex, Cloacal anomaly)	9	0.5
G25 Other specified and unspecified congenital malformations of musculoskeletal system	5	0.3

Diagnostic Grouping	Number of cases	Prevalence / 10,000
H. Other Congenital Anomalies (Q80 – Q86 & Q89)	**6**	**0.3**
H01 Icthyosis (Q80.8)	0	0
H02 Simian crease (82.8)	0	0
H03 All other congenital malformations not elsewhere classified (Q81.8, Q82.3, Q84.8, Q89.1, Q89.2)	6	0.3

Diagnostic Grouping	Number of cases	Prevalence / 10,000
I. Multisystem Anomalies / Syndromes	**35**	**1.9**
I01 Meckel Gruber Syndrome (Q61.9)	4	0.2
I02 Pierre Robin syndrome (Q87.0)	18	1.0
I03 Sirenomelia sequence (Q87.2)	4	0.2
I04 VACTREL (Q87.2)	0	0
I06 Other Syndromes (Q75.1, Q87.1, Q87.3, Q87.5, Q87.8, Q87.9)	9	0.5

Diagnostic Grouping	Number of cases	Prevalence / 10,000
J. Chromosomal Anomalies (Q90)	**34**	**1.8**
J01 Down's Syndrome (Q90.0 – Q90.9)	21	1.1
J02 Edwards' Syndrome (Q91.3)	6	0.3
J03 Patau's syndrome (Q91.7)	4	0.2
J03 Turner's Syndrome (Q96.0)	3	0.2

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