

Recognising Impairments at Birth

A Manual for Maternity Unit Personnel

Part of the CBM Prevention Toolkit
on Birth Impairments

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Contents

The CBM Strategy on Structural Birth Impairments	2
I. Primary Health and Birth impairments.....	4
A. Can birth impairments be prevented?	4
B. Preconception prevention of birth impairments:	5
C. Pre-term birth (and relationship to childhood impairments)	5
D. Post-conception pregnancy care:	7
E. The relationship of birth impairments to physical impairment.....	7
F. The relationship of birth impairments to visual impairment	7
G. The relationship of birth impairments to hearing impairment	7
H. The relationship of birth impairments to cognitive impairment and community mental health.....	8
II. Identification and Early Intervention for Structural Birth Impairments. 8	
Strategy for birth impairment intervention.....	9
1. Awareness, early identification and referral.....	9
2. Birth impairment surveillance programs	9
3. Advocacy.....	9
4. Curriculum saturation	10
5. Surgical intervention:.....	10
Brief description of impairments represented in the flipchart:	12
1. Oculo-cutaneous albinism:.....	12
2. Hydrocephalus	12
3. Oro-facial clefts (Cleft lip & palate)	13
4. Brachial Plexus Palsy.....	13
5. Congenital cataract.....	14
6. Impairments of the fingers & toes: Polydactyly / syndactyly	14
7. Impairments of the limbs – missing or deformed arm & leg parts .	14
8. Clubfeet (Congenital Talipes Equino-Varus, CTEV)	15
9. Developmental dislocation of the hip (DDH)	15
10. Neural tube defects (Spina bifida / myelodysplasia)	16
11. Impairments of the abdomen, genitals and anus:	16
Examining newborns for congenital impairments	18
References:	19

Preventing and Recognizing Impairments at Birth

Birth impairments are a common cause of mortality and disability in children in developing countries, yet there is little recognition or profile.

Each year at least 7 million children are born with serious birth impairments. The incidence of birth impairments is between 40 and 60 per thousand live births (4-6%), with a higher incidence in lower and middle income countries.

CBM's worldwide work assisting children and young adults with disabilities include many who experience disability as a result of birth impairments such as congenital cataracts, clubfeet, cleft lip and palate, and hydrocephalus.

Prevention, treatment and rehabilitation is possible for 70% of birth impairments in children.

CBM's initiative on prevention of disability from birth impairments will adopt two strategies:

- I. Primary prevention through effective pre-conception and maternal care
- II. Early identification and referral for structural birth impairments

The CBM Strategy on Structural Birth Impairments

- The CBM strategy on birth defects seeks to advocate for the recommendations of the WHO and The March of Dimes *Global Report on Birth Defects*¹, which follows.
- The Prevention of Disability "Toolkit" on **Primary Health** titled "How to Improve the Health and Development of your Child: Preventing Impairment & Disability" seeks to implement recommendations 1, 2 & 3 through educating mothers on pre-conception care, safe motherhood, good nutrition and timing of births.
- The Prevention of Disability "Toolkit" titled "**Recognizing Impairments at Birth**" seeks to implement recommendations 6 & 7; to train health professionals and birth attendants in early identification of birth defects, appropriate examination of newborns and early referral for treatment.

Recommendations of the expert group on birth defects, March of Dimes (2006):

1. Educate the community, health professionals and workers, policy makers, the media, and other stakeholders about birth defects and the opportunities for effective care and prevention.
2. Promote family planning, allowing couples to space pregnancies, plan family size, and provide information about the risks of giving birth before the age of 18 and after the age of 35.
3. Ensure a healthy, balanced diet during a woman's reproductive years through an adequate intake of macronutrients and a broad range of micronutrients, specifically iron and iodine. Add 400 micrograms synthetic folic acid to the diet by supplementation or fortification.
4. Control infections in all women of reproductive age.
5. Control chronic diseases, specifically insulin dependent diabetes mellitus, heart disease and epilepsy.
6. Train health professionals to recognize birth defects.
7. Conduct physical examination of all newborns before hospital discharge.
8. Advocate for the establishment of appropriate child health services to care for infants with birth defects.
9. Advocate for the establishment of national capacity for surveillance and monitoring of common birth defects
10. Support lay organizations including patients/parent support groups to advocate and educate their communities.

What are Birth Impairments?

Birth impairments can broadly be categorized into:

- Congenital malformations (structural birth defects)
- Chromosomal conditions (e.g. Down syndrome)
- Single gene defects (mainly cause enzyme defects, or inborn errors of metabolism)

Chromosomal conditions and single gene defects constitute a large and complex group of disorders. Many cause enzyme or metabolic abnormalities that are not obvious at birth except by biochemical evaluation. Few of these complex and expensive chemical studies are available in the developing world. Many of these children require complex medical management, but rarely surgical management. Many result in cognitive and developmental impairments.

Structural birth impairments, however, can often be seen with the naked eye and can be recognized at birth. Yet all too often birth attendants in busy

maternity units, or traditional birth attendants in the village, do not know how to recognize common birth impairments and are unaware of how to give appropriate advice to mothers. The result is an unnecessary i.e. preventable disability.

The flipchart "Recognizing Impairments at Birth" is intended as an educational tool for maternity workers.

But what can be done to prevent birth impairments in the first place? There are some effective strategies that can be implemented through community awareness, education and primary health practices. The flipchart "How to Improve the Health and Development of your Child: Preventing Impairment & Disability" has been created for this purpose as well as for integration into primary health programs.

I. Primary Health and Birth impairments

A. Can birth impairments be prevented?

At least 8 conditions may lead to a higher incidence of birth impairments:

1. Inadequate pre-conception intake of folic acid
2. Iodine deficiency in the mother's diet
3. Lack of vaccination against rubella (German measles)
4. Women giving birth after 35 years of age
5. Consanguineous marriages
6. Alcohol consumption during pregnancy
7. The use of teratogenic medications and environmental pollutants
8. Lack of prenatal genetic counseling

These then become the basis of a prevention policy.

There are effective prevention strategies for certain impairments, notably for the following:

- Spina bifida – preconception folate supplementation
- Rubella sequence – rubella vaccination
- Fetal alcohol syndrome – abstinence from alcohol before and during pregnancy
- Congenital hypothyroidism– Iodine supplementation

There is some evidence that multivitamin supplementation, including folate, during conception and early pregnancy can reduce the incidence of many structural birth defects.

There is also evidence that good nutritional support to all mothers results in healthier babies and less problems with developmental and cognitive delay.

B. Preconception prevention of birth impairments:

"Strategies for improving the health of women, mothers, newborns and children are essential for effective prevention and care of those with birth defects.

Effective interventions—including family planning, optimizing women's diets, managing maternal health problems and avoiding maternal infections – are feasible and affordable even in resource-poor environments."

March of Dimes executive report 2006

The following basic principles should be encouraged:

1. Basic reproductive education and health services.
2. Close relatives should not marry.
3. Provide information about the risks of giving birth after the age of 35
4. 400 micrograms folic acid per day pre and post-conception
5. Use iodized salt
6. Women should be vaccinated against Rubella and Tetanus before reproductive age
7. Control of chronic health conditions, notably anaemia, diabetes, obesity and hypertension
8. Do not smoke or drink alcohol

C. Pre-term birth (and relationship to childhood impairments)

Pre-term birth (prematurity) has a significant impact on mortality and impairment in babies and there is emerging evidence that pre-conception and post-conception care can minimize pre-term births. There is a synergy between prevention of birth impairments, prevention of pre-term birth and prevention of disabling impairments.

The definition of pre-term birth is birth before 37 weeks gestation.

Approximately 12 million babies are born prematurely each year.

29% of global neonatal mortality is related to pre-term birth (almost 1 million babies annually)

However, the impact is exacerbated by the fact that the other common causes of neonatal mortality, asphyxia and neonatal infections, are also related to prematurity.

The relationship of pre-term birth to cerebral palsy:

Premature babies are vulnerable to intra-ventricular bleeds into the brain, birth asphyxia and neonatal infections, all of which lead to brain damage and cerebral palsy.

The earlier the pre-term birth the greater the incidence of cerebral palsy and neurodevelopmental impairment in babies.

The relationship of pre-term birth to visual impairment:

Retinopathy of prematurity is caused by oxygen therapy for premature babies and is increasing in prevalence in middle income countries as newborn care expands.

Prematurity increases the risk of infections including ophthalmia neonatorum

The relationship of pre-term birth to hearing impairment

There is an increased incidence of hearing impairment in pre-term babies

The relationship of pre-term birth to cognitive impairment and community mental health

The earlier the pre-term birth the more the impact on the vulnerable developing brain of an infant, causing learning and cognitive developmental delays and epilepsy.

Risk factors:

Scientifically proven factors that increase the risk of pre-term birth are:

- Smoking
- Low body mass index (BMI) (inadequate nutrition)
- Hypertension
- Previous pre-term birth
- There is some evidence that stress, depression and partner violence may increase the risk of pre-term birth

Community Interventions that can reduce the impact of pre-term births and birth complications (pre-eclampsia), and hence birth impairment include:

- Appropriate nutrition
- Screening for maternal health and control of chronic diseases, specifically anaemia, diabetes, obesity and hypertension
- Control of maternal infections

The CBM prevention of disability “toolkit” on Primary Health Care includes information to improve pre-conception care at community level through

education of women and adolescent girls.

D. Post-conception pregnancy care:

The following basic principles should be encouraged:

1. Get pre-natal care at least 4 times during the pregnancy
2. Deliver in the presence of a skilled birth attendant
3. Avoid drinking alcohol during pregnancy
4. Do not smoke or use street drugs during pregnancy
5. Know HIV status and get appropriate care if positive
6. Get immediate medical attention for fevers
7. Avoid medications during pregnancy where possible. Medications during pregnancy should only be taken on a qualified doctors advice
8. Use iodized salt
9. Continue 400 micrograms of folic acid daily
10. Eat a well balanced nutritious diet rich in vitamins, minerals and protein. Attention to adequate calcium intake to ensure healthy fetal bone structure and healthy lactation.

E. The relationship of birth impairments to physical impairment

A large proportion of visible structural birth impairments result in limitations of physical functioning, including such conditions as clubfeet, arthrogryposis, oro-facial clefts and spina bifida.

F. The relationship of birth impairments to visual impairment

Congenital cataract, ophthalmia neonatorum and retinopathy of prematurity are birth related impairments that impact visual acuity.

G. The relationship of birth impairments to hearing impairment

Most hearing impairments cannot be seen with the naked eye and must be examined with special techniques and instrumentation. A common preventable cause of neonatal sensori-neural hearing loss is caused by inappropriate administration of aminoglycoside antibiotics, such as gentamycin. Rubella sequence, preventable by immunization, is a potent cause of hearing impairment in babies.

H. The relationship of birth impairments to cognitive impairment and community mental health

Many birth impairments result not just in limitations of physical and sensory functioning, but in cognitive impairment and developmental delay:

1. Pre-term birth is related to an increased incidence of cerebral palsy, which is a brain injury that can result in cognitive impairment and epilepsy
2. The common chromosomal defect Trisomy 21 results in Down syndrome which is accompanied by cognitive impairment
3. Many single gene defects result in children with syndromes of cognitive impairment.

Cerebral palsy is an example of a common birth impairment that can impact all of physical, visual, hearing and cognitive functioning.

Many structural birth impairments cannot be prevented and the strategy then must become secondary prevention through early identification, treatment and rehabilitation.

II. Identification and Early Intervention for Structural Birth Impairments.

Congenital Malformations are also called *Structural* Birth Impairments. These may or may not have a genetic basis but result in a structural change in the body which can usually be identified on physical examination. This is the group of birth defects that most impacts CBM's childhood disability strategy.

The most common structural birth impairments are:

1. Malformations of the brain and neurologic system
2. Malformations of the limbs
3. Heart defects.

Incidence:

Structural birth impairments occur approximately 30 per 1000 live births (WHO).

- About 25% are very severe resulting early death.
- About 50% are treatable or correctable.
- About 25% result in long-term disabilities despite the best of treatment.

Structural birth impairments almost invariably require surgery or surgical expertise for treatment. Where that surgical expertise is not available, as is

the case in most resource-poor countries, there is an increased risk of lifelong impairment and disability.

Prevalence statistics from our study in Rwanda show 23% of all musculoskeletal physical impairments in children are due to congenital malformations (structural birth impairments).

Congenital impairments seen in our medical projects include:

- Clubfoot
- Oro-facial clefts (cleft lip and palate)
- Polydactyly and syndactyly (extra digits or joined digits)
- Limb reduction deformities (absent, shortened, or malformed limbs)
- Neural tube defects (Spina bifida)
- Hydrocephalus

Medical eye care: Congenital cataract

Strategy for birth impairment intervention

1. Awareness, early identification and referral

Since many of birth impairments cannot be prevented primarily, disabling effects must be prevented by early intervention and treatment. This means referral to a specialist surgical service in almost all cases.

2. Birth impairment surveillance programs

Much work has been done on early identification of clubfeet and oro-facial clefts by training of primary health personnel and birth attendants. While very important, this has meant identification of only a limited percentage of all structural birth impairments. It is more appropriate to introduce birth impairment surveillance programs that create awareness and identification of *all* structural birth impairments. The flipchart toolkit "Recognizing Impairments at Birth" has been developed for training of primary health workers and birth attendants in comprehensive birth impairment surveillance.

3. Advocacy

Every baby should be examined carefully by a trained birth attendant before discharge from the maternity unit. Instruction in birth impairment

identification should be included in the curriculum of health training institutions. The flipchart toolkit "Recognizing Impairments at Birth" can be recommended and provided to training institutions. The toolkit contains an examination checklist for the common birth impairments.

Advocacy for effective referral pathways between local and district health services and specialized surgical and rehabilitation centers is encouraged.

4. Curriculum saturation

The Uganda Sustainable Clubfoot Care Project (USCCP) has shown the effectiveness of curriculum saturation of a specific birth impairment. The USCCP developed a curriculum and training module that could be applied to every level of healthcare education in the entire country including physicians, specialists, medical assistants, midwives, and primary health personnel. Thus all health-care personnel at every level of service delivery are aware of clubfoot deformity and the appropriate referral and treatment pathways. This successful model needs to be expanded to other areas of curriculum development in early identification and referral of birth impairments, as well as prevention strategies for other disabilities.

5. Surgical intervention:

Structural birth impairments almost invariably require surgery or surgical expertise for treatment. Where that surgical expertise is not available, as is the case in most resource poor countries, there is a higher risk of lifelong disability.

Early intervention is crucial to prevent permanent disability from many birth impairments. For example:

- If congenital cataract is treated early, normal vision is possible. If delayed, permanent blindness results.
- If shunting is delayed for hydrocephalus then this can result in permanent brain damage
- If clubfoot is identified early, it can be treated in the community with good results. If identified late, complex reconstructive surgery is required and the result is not as good.
- If cleft lip and cleft palate is not treated by surgery early, death from malnutrition is common.

The CBM strategy, therefore, needs to involve support for service delivery programs for these impairments.

Evaluating available health resources and developing referral pathways is necessary.

Some CBM partners have specialty surgical expertise. National referral hospitals should be accessed as much as possible. Sometimes specialty surgical care has to await visiting teams.

Some of the required surgeries are fairly simple, for example surgery for polydactyly, and may be available in district hospitals. Many of the surgical reconstructions, however, are very complex and require sub-specialist surgical expertise. This is not usually available even in national referral hospitals. There is a role for subspecialty children's hospitals, or units within general hospitals, in implementing surgical care for birth impairments and in training national surgeons in this very sub-specialized area.

There will be the need to network paediatric general (abdominal) surgical services, currently not available in most CBM programs. This would be for such conditions as gastroschisis (abdominal wall defects), imperforate anus, congenital hernias etc.

Neurosurgical services would be necessary for the treatment of neural tube defects (spina bifida) and hydrocephalus.

A birth impairment surveillance program has implications for areas of interest in CBM apart from babies with physical impairment, notably congenital cataracts and neonatal ophthalmic disorders, congenital deafness, and early intervention for children with Down syndrome.

What are the common structural birth impairments?

The prevention flipchart "Recognizing Impairments at Birth" encourages examination of all newborns by birth attendants and brings awareness to commonly identified structural birth impairments. These include:

1. Oculo-cutaneous albinism (albino babies)
2. Hydrocephalus
3. Oro-facial clefts (Cleft lip & palate)
4. Brachial Plexus Palsy
5. Congenital cataract
6. Impairments of the fingers & toes: Polydactyly / syndactyly
7. Impairments of the limbs – missing or deformed arm & leg parts
8. Clubfeet
9. Developmental dislocation of the hip
10. Neural tube defects (Spina bifida / myelodysplasia)

11. Impairments of the genitals and anus: Imperforate anus, Hypospadias

Brief description of impairments represented in the flipchart:

1. Oculo-cutaneous albinism:

Defect in the production of melanin, a pigment found in the skin, hair and the retina of the eye.

Incidence: varies.

Most prevalent in sub-Saharan Africa. Averages 1/4000 births in Africa.

Obvious skin and hair coloration at birth

A potent source of superstition, social isolation, discrimination and abuse.

Vulnerability to skin cancers

Visual impairment due to lack of protective pigments in the eye

Intervention:

Awareness. Little to be done in infancy.

Protection from sun exposure: use sunglasses, wide-brimmed hats, sunscreen

Regular vision checks

Medical/surgical intervention for skin lesions

2. Hydrocephalus

Enlarged head due to buildup of the cerebrospinal fluid which normally bathes the brain

Incidence: 0.5-0.9/1000

May be present at birth, but more commonly occurs progressively during early infancy

Commonly follows neonatal infections (meningitis, encephalitis) which blocks flow of cerebrospinal fluid by scarring.

This causes pressure on the brain tissue with progressive brain damage and impairment.

Intervention:

Urgent surgical referral to a specialized hospital unit. Delay in referral results in permanent physical and cognitive impairment.

Fluid can be surgically shunted from the brain to the abdomen, where it is absorbed (Ventriculo-peritoneal shunt), or endoscopic surgery can open an internal shunt (ETV)

Shunt failures can occur with growth, so community follow-up is very necessary

3. Oro-facial clefts (Cleft lip & palate)

Malformations producing a gap in the upper lip, and/or the soft palate and roof of the mouth. Occurs when these structures fail to join in the midline.

Incidence: Varies. 1-2/1000 live births

Often an obvious facial disfigurement resulting in stigmatization.

Babies have impaired sucking and may succumb to starvation and malnourishment.

Abnormal upper jaw and tooth development usually occurs.

When the palate is involved, speech is impaired.

Intervention:

1. Surgical treatment is very effective. It should be recommended as early as possible. Specialist paediatric anaesthetic skills and equipment are necessary to carry out the surgery safely since the surgery is accomplished within the airway.

2. Dental and maxillo-facial reconstructive services are often needed after primary surgery to close the cleft.

3. Speech therapy services.

The NGO's *Smile Train*, *Operation Smile* and *Operation Rainbow*, amongst others, have specialized in providing cleft services in lower income countries.

4. Brachial Plexus Palsy

This is not a congenital birth defect but an injury occurring during the birthing process. The most common cause is shoulder dystocia, where the baby gets stuck in the birth canal when the shoulder gets hung up. Attempts to forcibly deliver the baby result in stretching of the brachial plexus, the nerves running between the neck and the arm, resulting in partial paralysis of the arm.

The injury may be mild, recovering quickly, or severe and permanent.

The baby does not move the upper part of its arm. The baby cannot flex the elbow. The hand and wrist move well.

Incidence: Varies depending on maternity skills and resources. There is a lower incidence where caesarian section services are available.

Intervention: Referral to physiotherapy. Range-of-motion exercises prevent joints from becoming stiff. Where there is permanent loss of function in the upper arm, remedial activities can be taught.

Surgical treatment to graft the injured nerves is complex and highly

technological. It is not a realistic expectation in lower resourced countries.

Occasionally surgery is beneficial in older children to transfer tendons about

the shoulder, or perform corrective rotation surgery (osteotomy) of the humerus.

5. Congenital cataract

Opacification of the lens.

Usually occurs in both eyes

Often occurs in association with other birth defects

Incidence: 0.5-1/1000 in developed countries. Incidence unknown in low resource countries, probably greater

Identification: May be seen with the naked eye, but is better seen by shining a light into the eye. Flashlight or ophthalmoscope. The red reflex is absent or the lens seen is as a white opacification.

Intervention: Referral to a specialist eye center. Early identification and referral is very important because permanent blindness results with delay (the neural visual pathways do not develop).

Paediatric eye services are not always available in ophthalmology units.

Surgical correction is possible but is complex and difficult.

Babies and young children require general anaesthesia for surgery whereas most adults can be treated with local anaesthesia.

6. Impairments of the fingers & toes: Polydactyly / syndactyly

These are the most common structural birth impairments.

Polydactyly: extra fingers or toes

Syndactyly: joined fingers or toes

Absent or deformed digits can also occur

They do not usually result in severe functional impairment, but may result in stigmatization.

Incidence: Common 2/1000+. More common in black people, 13/1000

Intervention: Surgical referral. This is not urgent. Surgical treatment is best when the child is older than walking age.

7. Impairments of the limbs – missing or deformed arm & leg parts

This is a very diverse group of impairments, often occurring in association with multiple medical problems. Some categories include:

- Congenital amputations (0.5/1000) Babies born with missing parts of the upper or lower limbs.
- Reduction defects: One limb shorter than the other due to a missing segment of bone.

- Multiple stiff joints. (Arthrogryposis) (1/3000).
- Angulatory deformity: Limbs deformed in shape

Intervention: Referral to a specialist children's orthopaedic unit.

Surgical reconstruction is usually required. Surgery is very complex requiring specialized skills, technology and infrastructure that may not be available in lower income countries. These conditions can be improved but not cured.

Surgical care is not urgent but is best accomplished before school age.

Prosthetic and orthotic services might be necessary.

Wheeled mobility devices and walking aids might be necessary.

8. Clubfoot (Congenital Talipes Equino-Varus, CTEV)

The most common congenital defect causing locomotor impairment. Most commonly an isolated condition (Idiopathic Clubfoot), but often accompanies other birth defects and congenital anomalies such as arthrogryposis and spina bifida (Syndromic Clubfoot).

Soft tissue contractures on the inner and plantar aspect of the foot pull the foot downward and inwards into a deformed position. Without treatment children can walk but walk on the sides or top of their feet, have pain, and cannot wear normal shoes.

Incidence: Varies between 0.6 per thousand in Chinese and 8 per thousand in Polynesians. Worldwide average is 1.2 per thousand live births (1/800).

Intervention: early identification and intervention. Intervention within the first two years of life with the Ponseti method of manipulation and casting results in a cure. Older age children may need additional surgery. Orthotics [braces] are almost always needed after treatment. Long-term follow-up is necessary to monitor for relapse.

Clubfoot programmes, implementing the Ponseti technique, have been very successful and are now identified as a "core programme" within CBM projects.

9. Developmental dislocation of the hip (DDH)

This birth impairment cannot be seen with the naked eye but must be examined. It is therefore the most difficult to identify and teach.

One or both hips are dislocated from the joint, or are unstable and dislocatable. Lack of identification results in a severe limp, permanent walking impairment and arthritis.

Incidence: Full dislocation 1/1000. Unstable 1/100. The incidence varies amongst cultures. It is rare in Africans, common in central Europeans and indigenous population of the Americas.

Examination: every newborn should have their hips examined carefully. The hips are gently moved inward (adducted) and outward (abducted) in a specific way (Barlow's maneuver) and a shift or "clunk" is felt. Learning this maneuver requires specific training.

Prevention: might be possible by advocating and adopting the African carrying position with the baby carried and nursed with its legs around the parents waist and back (abducted), not on its side (legs adducted), for the first few months of life. Tight swaddling of the legs must be discouraged.

Intervention: A simple brace, keeping the legs abducted to the side, is effective but requires specific orthotic skills to apply appropriately. Usually worn for 3 months.

Early treatment results in a cure. Treatment after neonatal age requires specialized surgery.

10. Neural tube defects (Spina bifida / myelodysplasia)

The lower part of the spine and spinal cord is deformed. The spinal bones are not properly joined in the midline. Spinal cord tissue may be exposed to the outside with an open sore, or bulging soft tissue. Large hairy patches or other skin blemishes in the lower spine may indicate underlying structural damage. Hydrocephalus almost always accompanies this condition.

Incidence: Varies greatly. Highest incidence is in Chinese children: 6/1000

Prevention: Folic acid (folate) supplementation **before** conception reduces the incidence and severity drastically. Taking folate after identification of pregnancy is too late to prevent this impairment. Advocacy is therefore necessary for all women of childbearing age to receive supplementation. In some countries food sources have folate fortification.

Intervention: Urgent referral to specialized medical centers where neurosurgery is available. The skin defect needs to be surgically closed and the hydrocephalus shunted. Lack of urgent care results in a very high rate of mortality.

The condition cannot be cured and permanent impairment is invariable. Long term rehabilitation in a multi-disciplinary setting is required including orthotic and wheeled mobility services. Pressure sores and problems with bowel and bladder function are common. Specialized seating reduces the risk of pressure sores.

11. Impairments of the abdomen, genitals and anus:

This group of impairments invariably requires the services of paediatric general (abdominal) surgery and specialist paediatric anaesthesia. These

services are rarely available at district hospitals. Since some of these conditions are life threatening, requiring emergency referral, there is a high mortality in rural settings.

Intervention: Know and establish referral pathways for paediatric general surgery care

11.1 Gastroschisis

Abdominal contents (bowel) protrudes through an opening on the abdomen

Very dramatic birth presentation.

Almost always fatal unless immediate surgery is performed

The results of surgery are good

Refer to a paediatric general surgery facility

Immediate care is to cover the bowel with plastic food wrap

11.2 Bladder extrophy

The bladder exits through an abnormal opening on the abdominal wall, leaking urine

Not immediately life threatening

Refer for surgical reconstruction by paediatric urologist/general surgeon

11.3 Imperforate anus

There is no opening at the anus. The bowel inside is usually normal.

Often occurs in association with other birth defects

Urgent surgery to open the anus or perform a bypass colostomy is necessary

11.4 Hypospadias

The urethra exits above or below the end of the penis

Not an emergency, but should be surgically corrected early in life

11.5 Testicular swelling: Congenital Hernia and Hydrocoele

Testicular swelling from bowel loops herniating into scrotum.

May not be seen at birth but become evident in early infancy

Not an emergency, but should be surgically corrected to avoid obstruction and strangulation of the bowel, which is then life threatening

11.6 Ambiguous genitalia

It is unclear whether the baby is male or female; it appears that the penis is very small, or the clitoris too big

Urinary function is usually normal

These are complex cases from a surgical and ethical viewpoint, requiring sub-specialist referral

Surgical correction is possible early in life

Examining newborns for congenital impairments

The checklist for examining a baby for structural birth impairments should include:

1. Look at the baby's colour, skin and temperature – are they all normal?
2. Is the baby's head very big or very small ?
3. Does the face look normal ? Look especially at the ears and eyes.
4. Does the neck move fully?
5. Shine a light in the baby's eyes – does the pupil look white or cloudy?
6. Check for a cleft lip. Put your sterile finger inside the baby's mouth to check for cleft palate.
7. Look at the baby's arms and legs – do they look normal? Do the joints have normal range of motion?
8. Are there five fingers and five toes?
9. Feel the baby's arms and legs – are they strong?
10. Are the limbs flexible? Do the joints have a normal range of movement
11. Does the baby have clubfeet?
12. Look at the baby's back – are there any sores, marks or hairy patches on the spine?
13. Look at the baby's anus and genitals – is the hole for urine in the correct place? Are there swellings in the testicles?
14. Is the anus normal?

Expert opinion

Expert opinion on the prevention and care of birth impairments can be obtained from:

- **March of Dimes** (MoD).
This NGO has developed a focus on birth defects and has become a leading advocate on the subject. MoD background documents and annual reports contain a wealth of information on birth defects. MoD is a collaborating partner with the WHO.
- **The WHO** has a human genomics program and genomics resource center with primary focus on genetic research. Little is established on treatment resources. The WHO initiatives are supported by March of Dimes.
- WHO Factsheet on Congenital Anomalies
<http://www.who.int/mediacentre/factsheets/fs370/en/index.html>
- The **Centers for Disease Control and Prevention** (CDC) website has extensive resources
<http://www.cdc.gov/ncbddd/birthdefects/index.html>

- PHG Foundation, Foundation for Genomics and Population Health
<http://toolkit.bornhealthy.org/overview.pdf>

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