Recognising Impairments at Birth

FACT SHEET

Part of the CBM Prevention Toolkit on “Recognising Impairments at Birth”

September 2014

Contents

1. Strategies in Prevention of Disability from Birth Impairments

Children and young adults experiencing the disabling effects of birth impairments are commonly seen in CBM supported projects.

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

i. Primary prevention through effective maternal care
ii. Early identification and referral for structural birth impairments

Birth impairments (commonly referred to as "birth defects") are a common cause of mortality and disability in children, yet there is little recognition or profile.
Each year at least 7 million children are born with serious birth impairments.
The incidence of impairments is between 40 and 60 per thousand live births (4-6%), with a higher incidence in lower and middle income countries.

2. Categories of impairments at birth

Birth impairments can broadly be categorized into:

- Congenital malformations (structural birth impairments)
- Chromosomal disorders (e.g. Down syndrome)
- Single gene defects (mainly cause enzyme defects, or inborn errors of metabolism such as hemophilia and thalassemia)

Our early identification program will focus specifically on **structural impairments**, also called congenital abnormalities, which can be identified easily at birth by observation and basic physical examination. We will not focus on impairments which require sophisticated laboratory or other investigations to diagnose.

3. 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
4. Women giving birth after 35 years of age
5. Consanguineous marriages
6. Alcohol consumption during pregnancy
7. The use of teratogenic[1] medications and environmental pollutants
8. Lack of prenatal genetic counseling

These then become the basis of a prevention policy.

[1] Interrupting or altering the normal development of a foetus with evident results at birth

4. Pre-conception prevention of birth impairments

The most effective prevention strategy starts **before conception**.

Strategy: Effective preconception prevention of birth impairments is accomplished through a **primary health care** approach.
Community programs are encouraged to engage the community, primary health facilities, and maternity units with the following information:

1. Basic reproductive health services.
2. Close relatives should not marry.
3. Inform women about the risks of giving birth after the age of 35
4. Take 400 micrograms folic acid per day peri-conception
5. Use iodized salt
6. Women should be vaccinated against Rubella before reproductive age
7. Avoid alcohol, illicit drugs and smoking
8. Avoid medications during pregnancy where possible. Medications during pregnancy should only be taken on a qualified doctors advice
9. Know HIV, hepatitis and syphilis status and get treatment if positive
10. Get medical advice and control for chronic diseases, notably anaemia diabetes, obesity and hypertension.

The information community rehabilitation workers should know and teach regarding prevention of birth impairments is contained in the CBM Primary Health Manual and flip chart entitled "*How to Improve the Health and Development of your Child; Preventing Impairment and Disability*".

5. Post-conception and prenatal care

1. All of the above should continue to be encouraged.
2. Mothers should attend prenatal care at least 4 times during pregnancy.
3. Encourage good nutrition, including adequate iron intake.
4. Mothers should be encouraged to give birth in the company of a skilled birth attendant.

Good pre-conception and perinatal care reduces the risk of pre-term birth (prematurity), which has a significant impact on preventing birth impairments in babies and pregnancy related impairment in mothers.

6. Incidence of Structural Birth Impairments

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

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

Prevalence statistics from Africa indicate that 25% of all musculoskeletal impairments in children are due to structural birth impairments.

Many structural birth impairments cannot be prevented!
The **strategy** then becomes early identification, treatment and rehabilitation. Prevention, treatment and rehabilitation is possible for 70% of birth impairments.

7. Early identification and referral

Strategy: Awareness, early identification and referral targeting maternity workers, birthing units and medical education schools.

- A flip chart and manual have been developed with visuals to educate maternity workers to identify and refer babies with common structural birth impairments (see *Recognising Impairments at Birth*)
- The standard of care is that birth attendants examine carefully all newborns before discharge to identify structural birth impairments.
- The flip chart aids birth attendants and maternity workers to identify the common structural birth impairments.
- The flip chart may also be used to educate and create awareness in the community as deemed appropriate

Congenital conditions seen in CBM medical projects and covered in the package include:

- Occulo-cutaneous Albinism
- Hydrocephalus
- Oro-facial clefts (cleft lip and palate)
- Brachial plexus injury
- Congenital cataract
- Polydactyly and syndactyly (extra digits or joined digits)
- Limb reduction deformities (absent, shortened, or malformed limbs)
- Clubfoot
- Congenital Dislocation of the Hip
- Neural tube defects (Spina bifida)
- Genital malformations

8. Early Intervention for Structural Birth Impairments

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, lifelong disability can be expected.
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 palate is not treated by surgery early, death from malnutrition is common.

The CBM strategy should therefore involve support for service delivery programs for these impairments.
Clubfoot programmes, implementing the Ponseti technique, have been very successful and are now identified as a “core programme” within CBM projects.

9. 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 has potential to be expanded to other areas of curriculum development for early identification and referral of birth impairments, as well as prevention strategies for other disabilities.
CBM projects are encouraged to disseminate the prevention toolkit education materials as widely as possible within training institutions.

10. References:

1. March of Dimes Global report on Birth Defects. The hidden toll of dying and disabled children. 2006
2. Reducing Birth Defects. Meeting the challenge in the Developing World. Institute of Medicine. 2003
3. Ponseti Clubfoot Management, a manual for Healthcare providers in Uganda. Global-help.org

Recognising Impairments at Birth

A Manual for Maternity Unit Personnel

Part of the CBM Prevention Toolkit on Birth Impairments

September 2014

Contents

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.

<u>The relationship of pre-term birth to cerebral palsy:</u>

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. Clubfeet (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

References:

1. March of Dimes Global report on Birth Defects. The hidden toll of dying and disabled children. 2006
2. Reducing Birth Defects. Meeting the challenge in the Developing World. Institute of Medicine. 2003
3. *Disabled Village Children*, chapters 11, 12, 14, 18, 22
David Werner, Hesperian Foundation 2009.

RECOGNISING IMPAIRMENTS AT BIRTH



cbm
together we can do more

CBM is one of the world's largest international disability and development organisations, committed to improving the quality of life of persons with disabilities in the poorest countries of the world. Based on its Christian values and over 100 years of professional expertise, CBM addresses poverty as a cause and a consequence of disability, and works in partnership to create a society for all.

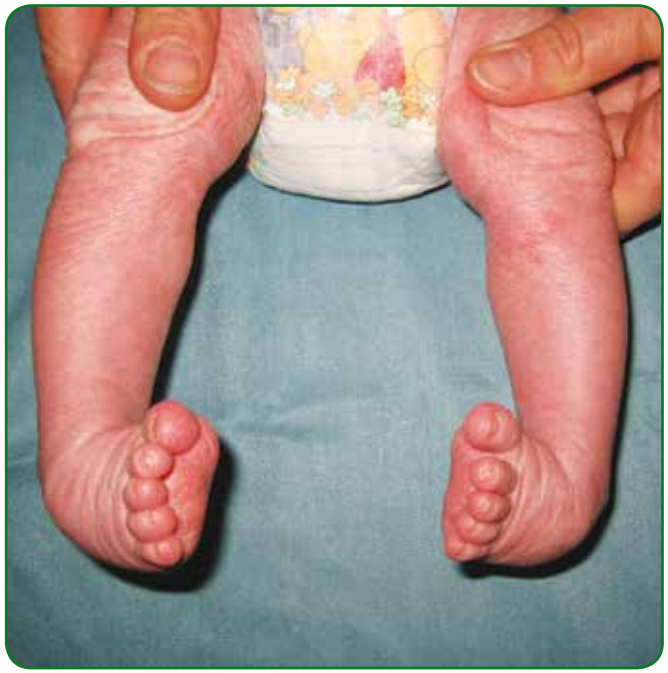
About impairments at birth

- They can happen to babies all around the world.
- They mostly occur when the baby is being formed very early in pregnancy.
- Impairments are NOT caused by witchcraft.
- They are NOT caused by a curse.
- They are NOT the fault of the mother and do NOT occur because the mother did something wrong.

About impairments at birth

Cleft Lip

Clubfoot



Cataract

Occulo-cutaneous albinism (Albino)

- The baby's skin has no pigment so it looks white.
- The hair also has no pigment and is white.
- Babies with albinism have problems with their eyes.
- Babies with albinism should have regular eye examinations.
- The skin of babies with albinism is damaged easily by the sun. All albino babies should be protected from the sun by wearing clothes and covering their heads.
- Babies with albinism have standard intelligence and grow like other children.
- Children with albinism have the same rights as other children to receive attention, food, schooling, and participate in play.

Occulo-cutaneous albinism (Albino)

Hydrocephalus

- Hydrocephalus causes the head to be too big and soft. Does the baby's head look too big?
- The eyes may look like the "setting sun".
- The baby should be taken immediately to a special hospital for surgery to insert a shunt to drain the excess fluid.
- Hydrocephalus should be treated urgently to prevent brain damage.
- The baby should be seen at a rehabilitation centre to get (physio)therapy.
- The nearest hospital for treating babies with hydrocephalus is _____.
- Children with hydrocephalus have the same rights as other children to receive attention, food, schooling, and participate in play.

Hydrocephalus

Baby with hydrocephalus
(photo: CURE International)

"Setting sun" sign: the eyes are directed downward
(photo: Bryce Flurie)

Baby after shunt operation

Happy child with controlled hydrocephalus

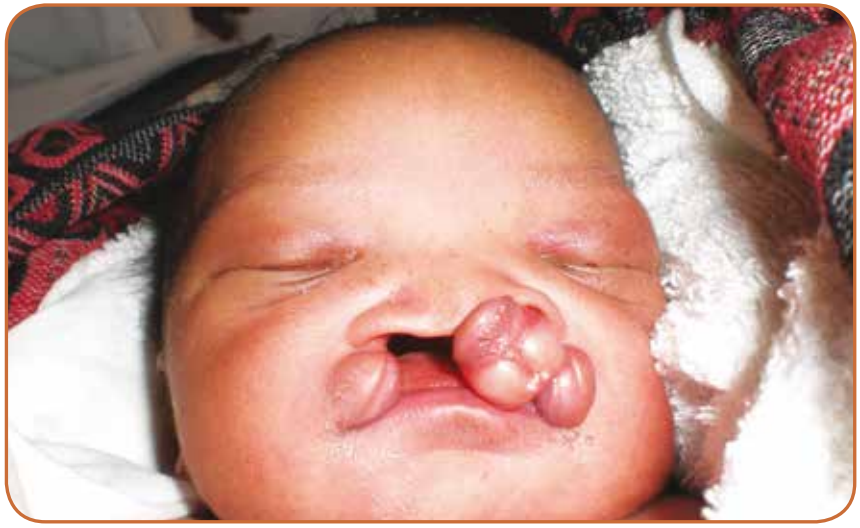
Cleft lip and palate

- Cleft lip/palate is a common impairment.
- A cleft is a gap in the skin of the upper lip that extends to the base of the nose.
- Sometimes the palate has a cleft but the lip is intact.
- Every newborn should be examined for cleft lip and palate.
- Look in the mouth and feel for cleft palate with a clean finger.
- Clefts can be repaired by surgery.
- Refer the baby as soon as possible to a specialised hospital performing this type of surgery.

Feeding

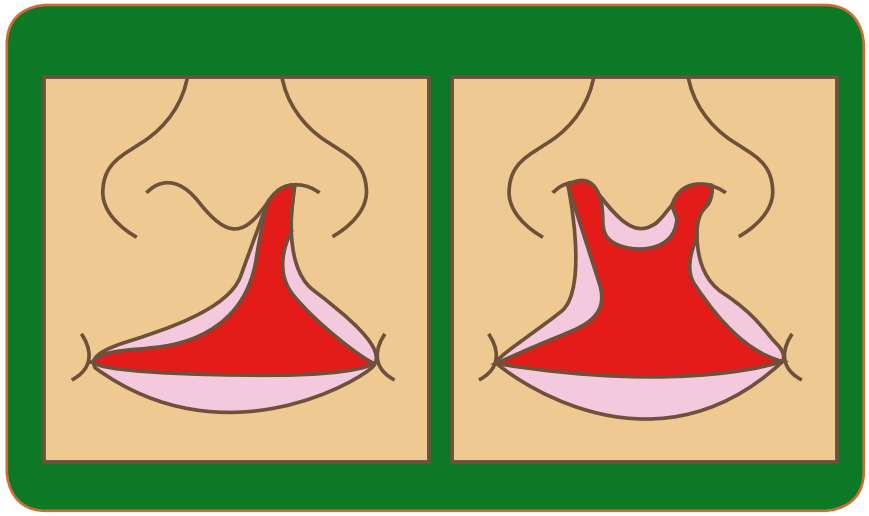
- Babies with cleft lip and palate have difficulty feeding.
- Breast milk is best.
- Feed more often per day.
- Keep the baby upright when feeding.
- If the baby has problems sucking, express breast milk and give it with a spoon.
- Wind/burp the baby more often.
- Give infant food or boiled cow's milk if there is not enough breast milk.

Cleft lip and palate

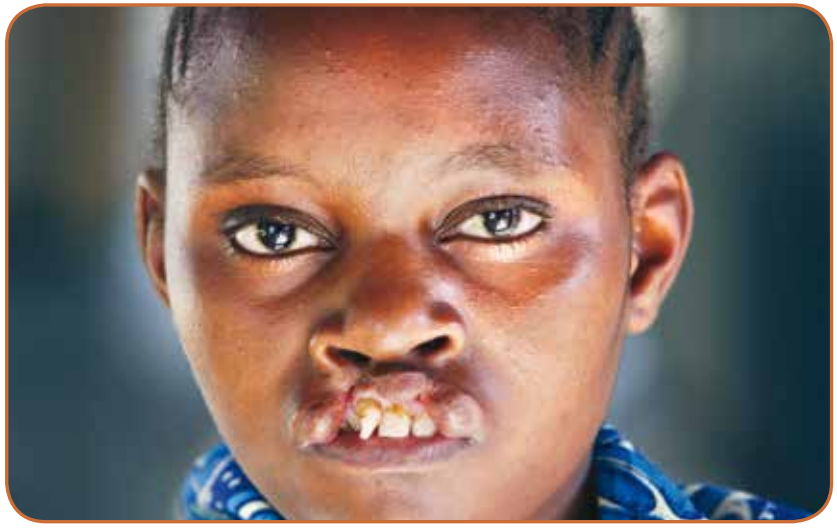


Cleft lip and palate before surgery

After surgery



A cleft can be one side or two



A child with untreated cleft lip

Brachial plexus injury

- Brachial plexus injury occurs during difficult delivery when the head or arm needs to be pulled hard.
- The nerves between the neck and the arm, the brachial plexus, become stretched too much causing partial paralysis of the arm.
- The baby holds one arm at the side, with the elbow straight and the fingers clenched.
- The arm does not move like the other one when the baby is stimulated.
- The hand may still move.
- Sometimes the baby seems in pain when moved during the first few days.
- Recovery often occurs, but it may take many months. Sometimes there is permanent impairment.
- Send the baby to a physiotherapist for passive movements and stimulation.
- Look carefully for brachial plexus injury in any baby who has had a difficult delivery.

Brachial plexus injury

The arm does not move like the other one when the baby is stimulated.

Cataract

- Check the baby's eyes.
- The pupil should look black.
- Shine a torch into the eye.
- If there is a cataract the pupil looks white or cloudy.
- Cataract can cause blindness.
- Refer the baby immediately to an eye hospital.
- The nearest eye hospital is _____.
- A small operation can cure cataract.

Cataract

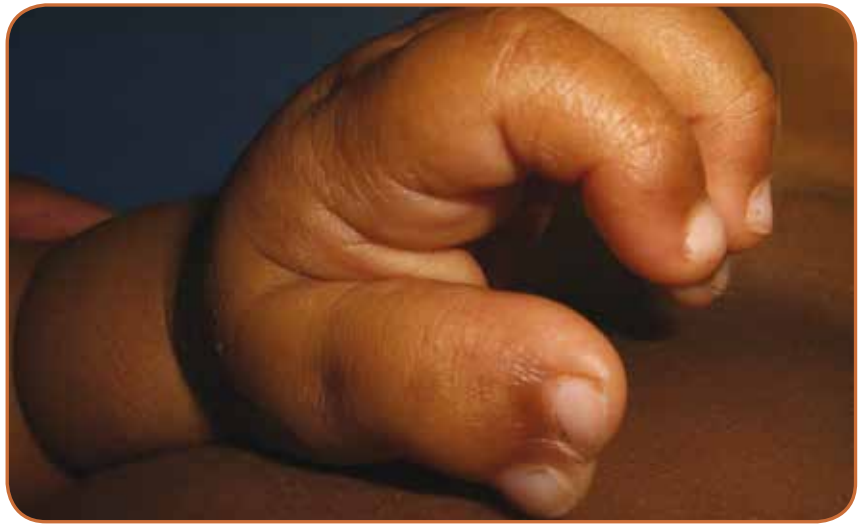
If the pupil looks white or cloudy it could be cataract

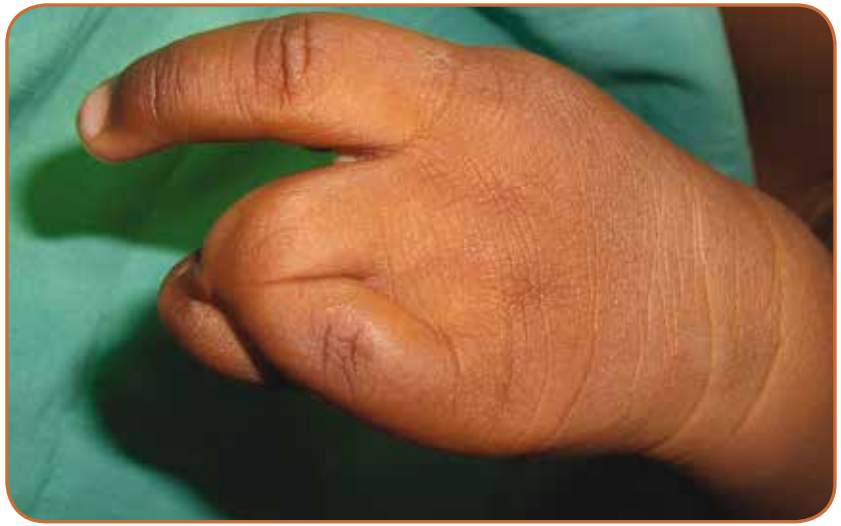
Check the eye with a torch

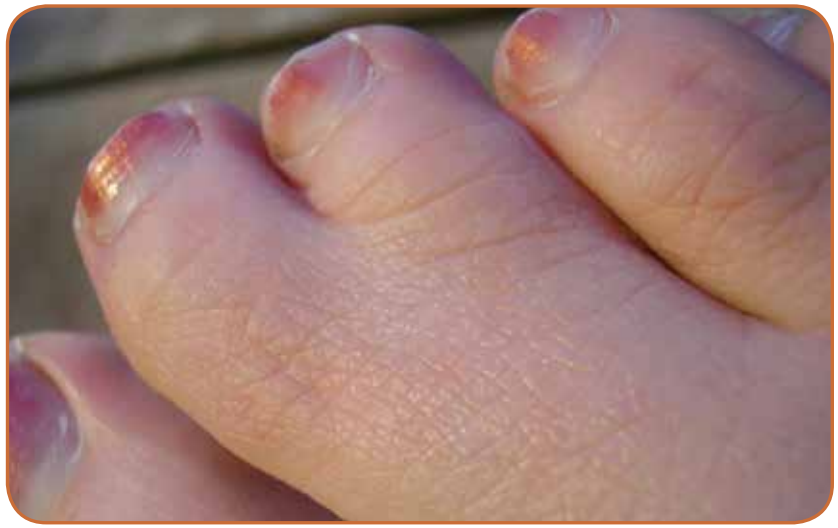
Impairments of the fingers and toes

- Are there the proper number of fingers and toes? An extra finger or toe is called “polydactyly”.
- Is the shape of the finger or toe normal?
- Are the fingers or toes joined together? This is called “syndactyly”.
- Most finger and toe impairments do not cause serious loss of function, but can cause embarrassment and result in discrimination.
- These impairments and loss of function can be improved by surgery.

Impairments of the fingers and toes



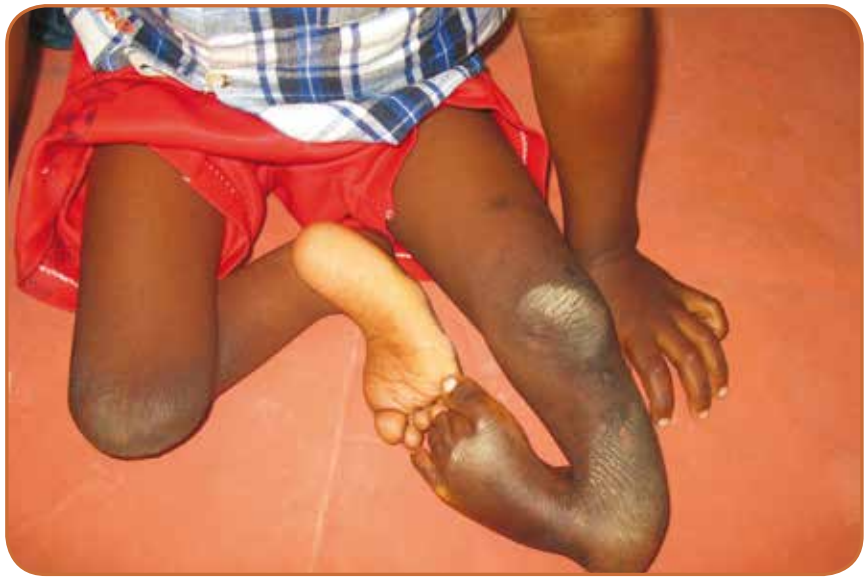




Limb impairments

- Babies can be born with part of a limb missing.
- The limb can be shortened or impaired.
- Do the baby's limbs move normally?
- Babies with a lot of joint stiffness have a condition known as arthrogryposis.
- Children with limb impairments can be helped at specialised rehabilitation centres.

Limb impairments

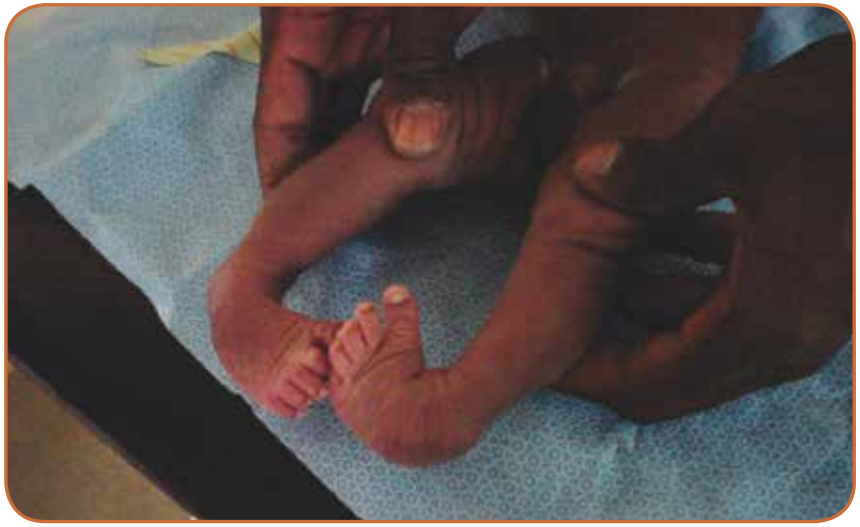


Arthrogryposis

Clubfoot

- Clubfoot can be treated.
- If it is left alone there is severe impairment and children walk on the side of their foot.
- Babies with clubfoot should be referred as soon as possible to a clubfoot clinic.
- The nearest clubfoot clinic is _____
- Treatment with casts can result in a normal foot and normal function.

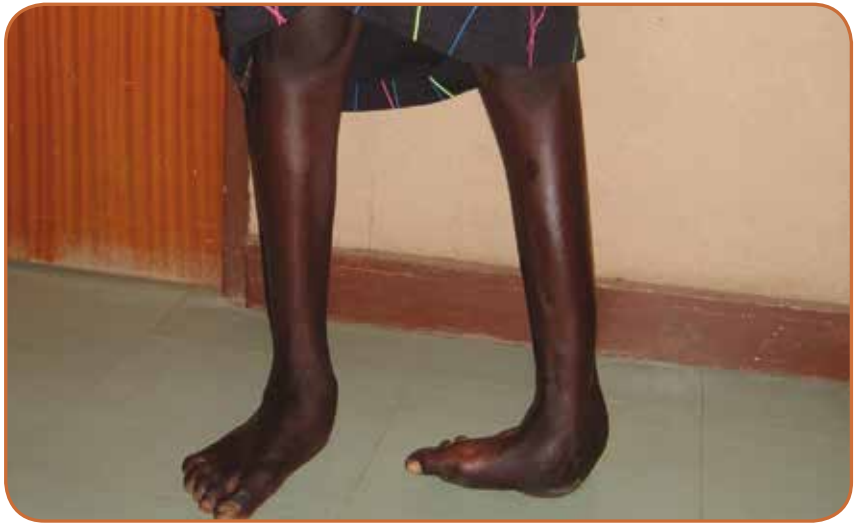
Clubfoot



Clubfoot

Treated with casts

Regular follow-up with special foot abductor brace is required

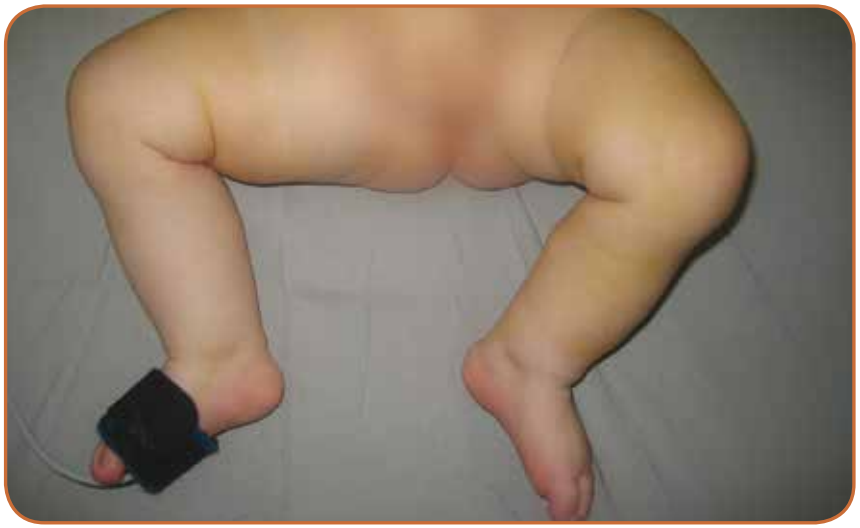


Untreated clubfoot leads to severe impairment

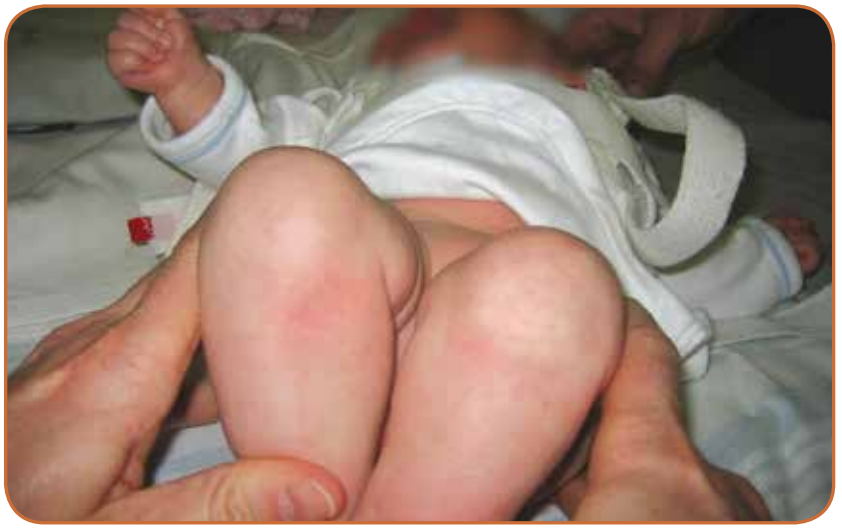
Dislocation (developmental) of the hip

- In this condition, the baby's hip joint is unstable and dislocates when the legs are brought together.
- It is more common in babies born breech.
- It is difficult to see. The leg may look shorter and have an extra crease.
- An examination is necessary for detection.
- The legs are first bent up to the bottom. Check if one looks shorter than the other.
- Then the legs are gently moved to the side. A shift or "clunk" is felt as the hip goes in and out of the joint.
- Carrying a baby on your back or side, Africa style, helps to correct hip dislocations.

Dislocation (developmental) of the hip



The left leg looks short and there is an extra crease.



First bring the legs together. Is one shorter than the other?

Then move the legs gently back and forwards.

Feel for the hip moving in and out of the joint.

Spina bifida

- Check the baby's back. If there is a large bulge, a sore, or even an unusual hairy patch, the baby has spina bifida.
- Spina bifida is a serious problem. It is caused when the baby's back does not close properly during development in the womb.
- There is damage to the spinal cord and there may be paralysis of the legs and incontinence.
- Often these babies also have hydrocephalus.
- Babies with spina bifida can die easily from infection.
- Treatment is very difficult.
- When the damage to the spinal cord is severe, the child needs to use a wheelchair.
- When the damage is mild, the child may still be able to walk with assistance (crutches and/or braces).
- The nearest hospital for treating babies with spina bifida is _____.

Spina bifida

Bulge on baby’s back

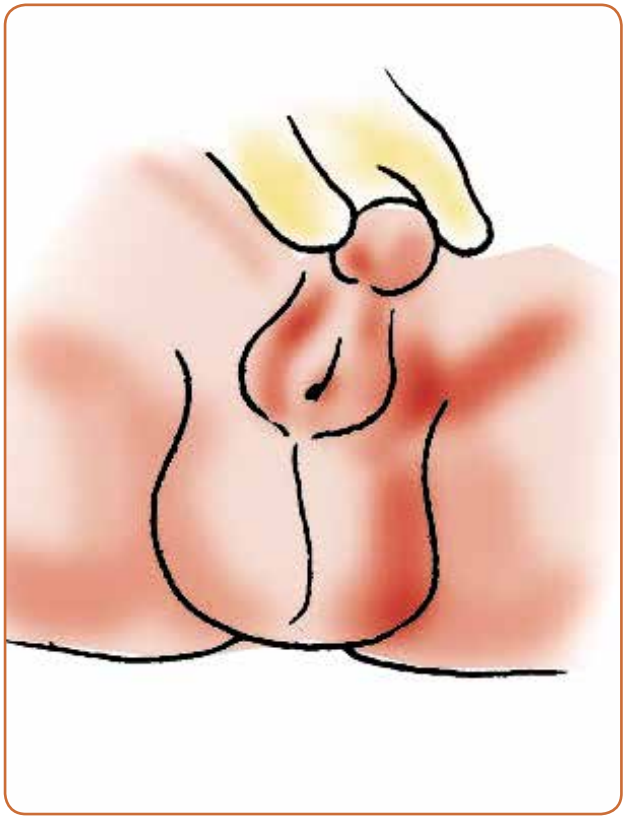
Problems of the genitals and anus

- Examine the genitals and anus of a newborn carefully.
- Do they look normal?
- Bladder exstrophy is where the hole for the urine is on the abdomen.
- Hypospadias is where the hole for urine (urethra) is below where it should be.
- If the testicles look large the baby may have a hernia or hydrocoele.
- These problems can be fixed by surgery.
- Is there a hole for the anus and has the baby passed meconium? Then, the baby may have an imperforate anus.
- Imperforate anus must be corrected by urgent surgery.
- Refer the child immediately to the referral hospital.

Problems of the genitals and anus

Bladder exstrophy

Imperforate anus

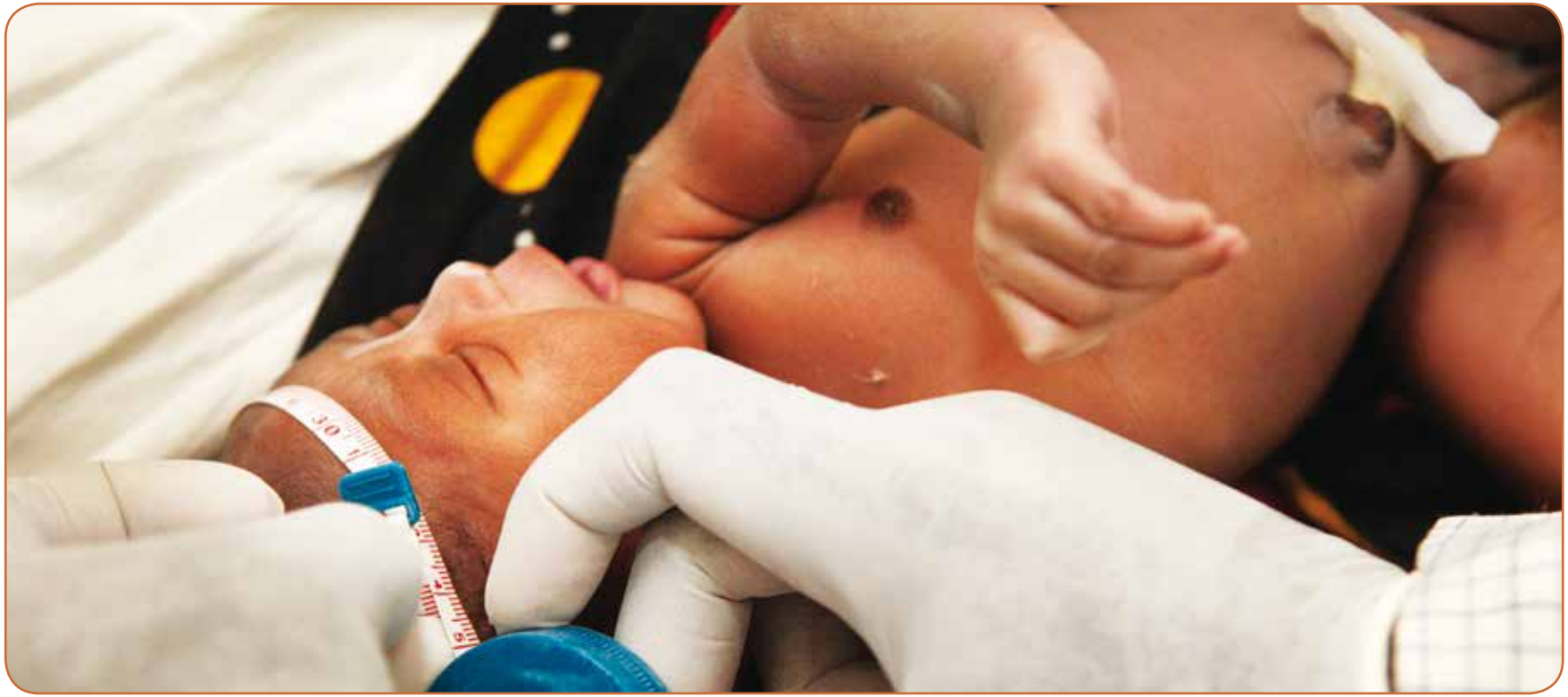


Hypospadias

Examining newborns for impairments

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

Examining newborns for impairments



RECOGNISING IMPAIRMENTS AT BIRTH

Index

This manual was produced in Tanzania by CCBRT and CBM with EU/CBM funding as a component of its joint programme with APDK, Kenya. This programme aims to reduce the prevalence of disabilities and mitigate the effects of impairments through improved maternal and newborn care and greater community awareness.

COMPREHENSIVE COMMUNITY BASED CCBRT REHABILITATION IN TANZANIA
Changing Lives, Changing Communities

apdk
Empowering persons with different abilities

cbm
together we can do more