सत्यमेव जयते

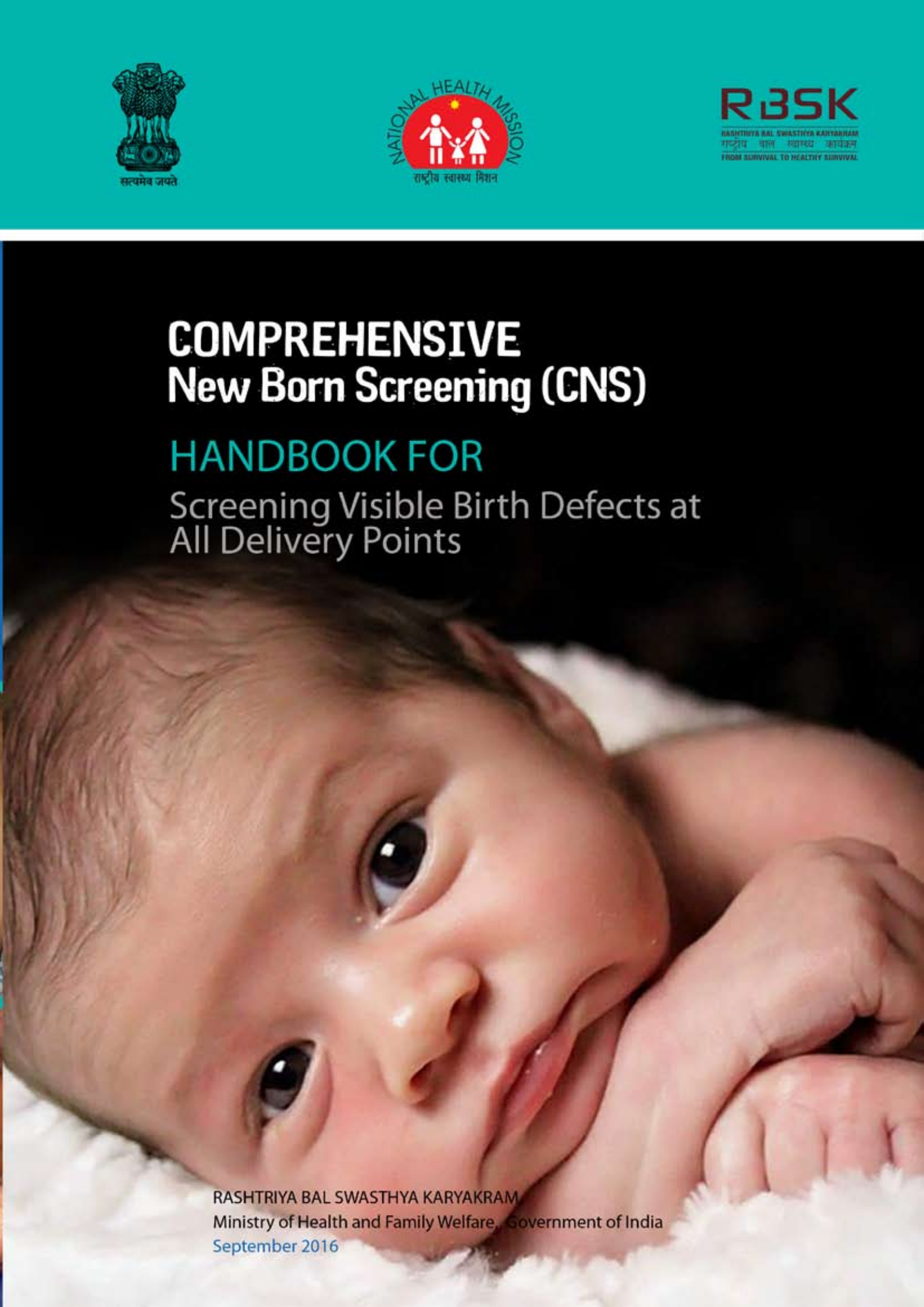
NATIONAL HEALTH MISSION
राष्ट्रीय स्वास्थ्य मिशन

RBSK
RASHTRIYA BAL SWASTHYA KARYAKRAM
राष्ट्रीय बाल स्वास्थ्य कार्यक्रम
FROM SURVIVAL TO HEALTHY SURVIVAL

COMPREHENSIVE New Born Screening (CNS)

HANDBOOK FOR

Screening Visible Birth Defects at All Delivery Points



RASHTRIYA BAL SWASTHYA KARYAKRAM
Ministry of Health and Family Welfare, Government of India
September 2016

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DISCLAIMER

The contents of this book are for the purpose of general information and training to public health care personnel and are not meant for any commercial use.

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ABOUT THE DOCUMENT

This handbook has been developed for field level functionaries, particularly ANM and Staff Nurses, positioned at delivery points, to serve as a ready reference for conducting screening of Visible Birth Defects and their reporting. The document comprises following essential areas:

1. Identification
2. Recording & reporting
3. Initial management
4. Counselling the parents

The key elements of the handbook are:

a. Poster template for Visible Birth Defects as a ready reckoner - to be displayed at all Delivery Points

b. Reporting format - to be printed and made available at all Delivery Points for reporting details of every child identified with Birth Defect

c. An Atlas on Common Birth Defects with ICD 10 codes to facilitate identification

This is first of the series of manuals for detection of Birth Defects and has been developed to facilitate **Newborn screening for visible Birth Defects** at all delivery points through physical examination by the existing staff viz., ANM/ Staff Nurses/ Medical officers.

Subsequent manuals will deal with:

a. *Functional defects* like Vision Impairment, Hearing impairment and Congenital Heart Defects identified with the aid of non-invasive Instruments.

b. *Metabolic defects* like Haemoglobinopathies and Inborn Error of Metabolism identified through blood examination.

c. *Neurodevelopmental defects* associated with complications of prematurity identified through neurological examination.

Dr. Arun Kr. Singh
National Advisor, RBSK

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सत्यमेव जयते

भारत सरकार
स्वास्थ्य एवं परिवार कल्याण मंत्रालय
स्वास्थ्य एवं परिवार कल्याण विभाग
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Government of India
Ministry of Health & Family Welfare
Department of Health & Family Welfare
Nirman Bhavan, New Delhi - 110011

FOREWORD

As per available estimates, 6% of children are born with serious Birth Defects every year worldwide. In our country, annually more than 17 lakh children are born with a Birth Defect contributing to 10% of neonatal mortality and 4% of infant mortality. Hence, it is imperative that we address this significant issue to improve the survival and health outcome of our children. The increase in institutional deliveries in India provides us a unique opportunity to screen the newborn at delivery points. This window of opportunity, if missed, could be either fatal for the child or lead to a permanent disability.

Ministry of Health & Family Welfare, Government of India is giving utmost priority to strategies for prevention and management of Birth Defects under RBSK to ensure better survival outcomes of our children. Newborn screening for identification of abnormal conditions is one of the critical interventions initiated under RBSK. Screening of newborns at delivery points is a crucial component of this strategy for which the healthcare personnel at delivery points including Doctors, Staff Nurses and ANMs need to be thoroughly oriented and trained enabling them to identify, record and refer the cases to appropriate centers. This handbook is especially aimed to guide the field level functionaries, the ANMs and Staff Nurses in identifying visible Birth Defects and capture the data in the RCH register.

I hope that States and UTs will take advantage of this initiative so that we together can secure and promote the health of our children.

(Arun Kumar Panda)

New Delhi
27th September, 2016

वन्दना गुरनानी,भा.प्र.से.
संयुक्त सचिव

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सत्यमेव जयते

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Preface

Birth Defects have been recognized globally as a major contributor to neonatal and infant mortality and disability. The current global estimate of 6% of children being born with Birth Defects annually is based on the Global Report on Birth Defects (2006). World Health Statistics Report (2012) states that 97.8 lakh children are born annually with a serious birth defect worldwide and 94% of these occur in low and middle income countries. As per WHO SEARO report (2013), in India 7% of under 5 year deaths are attributed to congenital anomalies or birth defects.

Being aware of the impact of Birth Defects on our future generation, Ministry of Health & Family Welfare, Government of India is giving utmost priority to strategies for their identification, prevention and management. Early identification with assured linkage to care, support and treatment introduces equitable child health care approach which in the long run will reduce the burden of disability, improve health and ensure development of children born with Birth Defects.

Comprehensive Newborn Screening (CNS) Handbook for screening visible birth defects has been developed as a tool to aid Staff Nurses and ANMs at all delivery points. The handbook contains all relevant information for identification of Birth Defects visible on physical examination, their initial management, documentation and referral procedure and counseling of parents.

I believe that States/UTs will give due importance to newborn screening under RBSK and making use of this handbook for the purpose will ensure its proper implementation as envisaged.

(Vandana Gurnani)

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Nirman Bhavan, New Delhi - 110011

Acknowledgement

The Rashtriya Bal Swasthya Karyakram aims at early identification and early intervention of 4D's - Defects at birth, Deficiencies, Diseases and Developmental delays including disabilities- in children from birth to 18 years of age. Early identification of Defects at birth is an important component of RBSK that in the long run reduces the burden of developmental delays and disabilities. Aiming to serve as a tool aid for screening of visible Birth Defects by field level functionaries, this handbook has been developed with inputs from experts around the country.

This is to acknowledge the contributions of Dr. Madhulika Kabra, Division of Genetics, AIIMS, New Delhi, Dr. Neeraj Gupta, AIIMS, New Delhi, Dr. Minu Bajpai, Pediatric Surgery, AIIMS, New Delhi, Dr. Seema Kapoor, MAMC, New Delhi, Dr. Sujata Sinha, Technical Consultant, NHM Uttarakhand, Dr. Arun Singh, National Advisor, RBSK, members of the Technical Resource Group and subject experts from across the country who helped in developing this handbook.

(Dr. Ajay Khera)

TABLE OF CONTENTS

1. INTRODUCTION

The term Birth Defect encompasses a diversity of health conditions including physical malformations such as cleft lip or palate, chromosomal abnormalities such as Down syndrome, functional defects including sensory deficits such as congenital deafness and congenital cataract, metabolic defects including inborn error of metabolism or Haemoglobinopathies, neurodevelopmental disorders, and complications related to prematurity[1]. Birth Defects usually manifest at birth and can be identified at the delivery points; however, some defects do not begin showing symptoms until a few weeks or months later.

Some Birth Defects are externally visible, while some are not and would require the help of instruments. In addition, some would require invasive methods like blood examination to identify any of the metabolic errors or any abnormality in the formation of hemoglobin.

Realising the importance of Birth Defects and its implications on the affected individual, family and society, Ministry of Health & Family Welfare, Government of India under **Rashtriya Bal Swasthya Karyakram (RBSK)** initiated **Newborn screening for Birth Defects at all delivery points** with special focus on visible Birth Defects. Those Newborns identified with Birth Defects will be referred to higher centres for further evaluation and management.

Under this initiative, a comprehensive clinical examination is to be performed on all babies, usually within the first 48 hours of life. This consists of a full physical assessment from head to toe - to identify any visible defects, and also to reassure parents that their baby is healthy. The examination is to be performed by the Staff Nurse/ANM, facilitated by the Medical Officer.

The Newborn examination is part of a range of post birth screening opportunities, which includes: a) a Staff Nurse check immediately after birth for visible Birth Defects b) hearing, vision testing c) neonatal blood spot examination and d) a further physical examination at follow up visits in high risk cases.

This handbook is meant for ANMs/Staff Nurse at all delivery points and focuses exclusively on **Newborn screening for visible Birth Defects**.

1 National Birth Defects Prevention Network, CDC

2. SCREENING METHODOLOGY

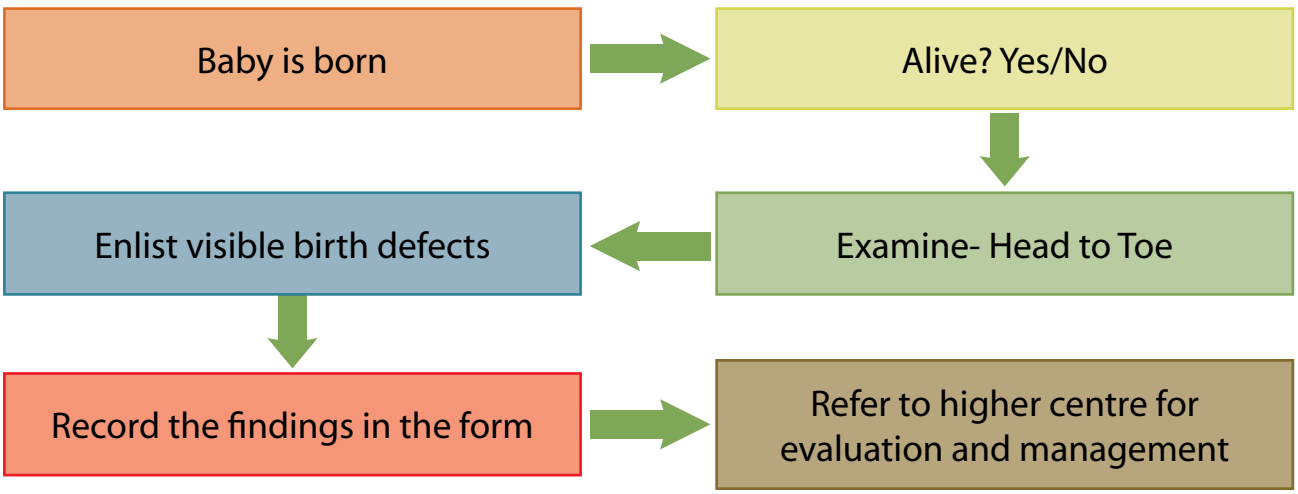
2.1 APPROACH TO NEWBORN SCREENING

Every Newborn requires a brief physical examination within the first few minutes after birth to assess the immediate vitals like respiration, heart rate etc., and then a full and detailed assessment for identifying Birth Defects, prior to discharge from the facility. High risk cases would require a follow up assessment for identifying neurodevelopmental delay. The physical examination component of the Newborn assessment is the most important step for identifying major congenital anomalies. There is no optimal time to detect all abnormalities; however, one should not miss the opportunity to screen at birth.

Use a systematic approach to examine the Newborn where possible. A recommended systematic approach is 'head to toe' and 'front to back'. Undress the Newborn down to the nappy as it is not possible to fully examine a dressed baby for all abnormalities.

Components	When and where	Who will do	How to do
Head to toe examination for visible birth defects	**a. At birth** **b. At all delivery points**	Existing ANM/ staff nurse and Doctors	Detected through proper physical examination of the new born– through head to toe examination using pictorial tool. For example, Neural Tube Defect, Cleft Lip Palate, Down Syndrome, Limb deformities, Club foot etc.

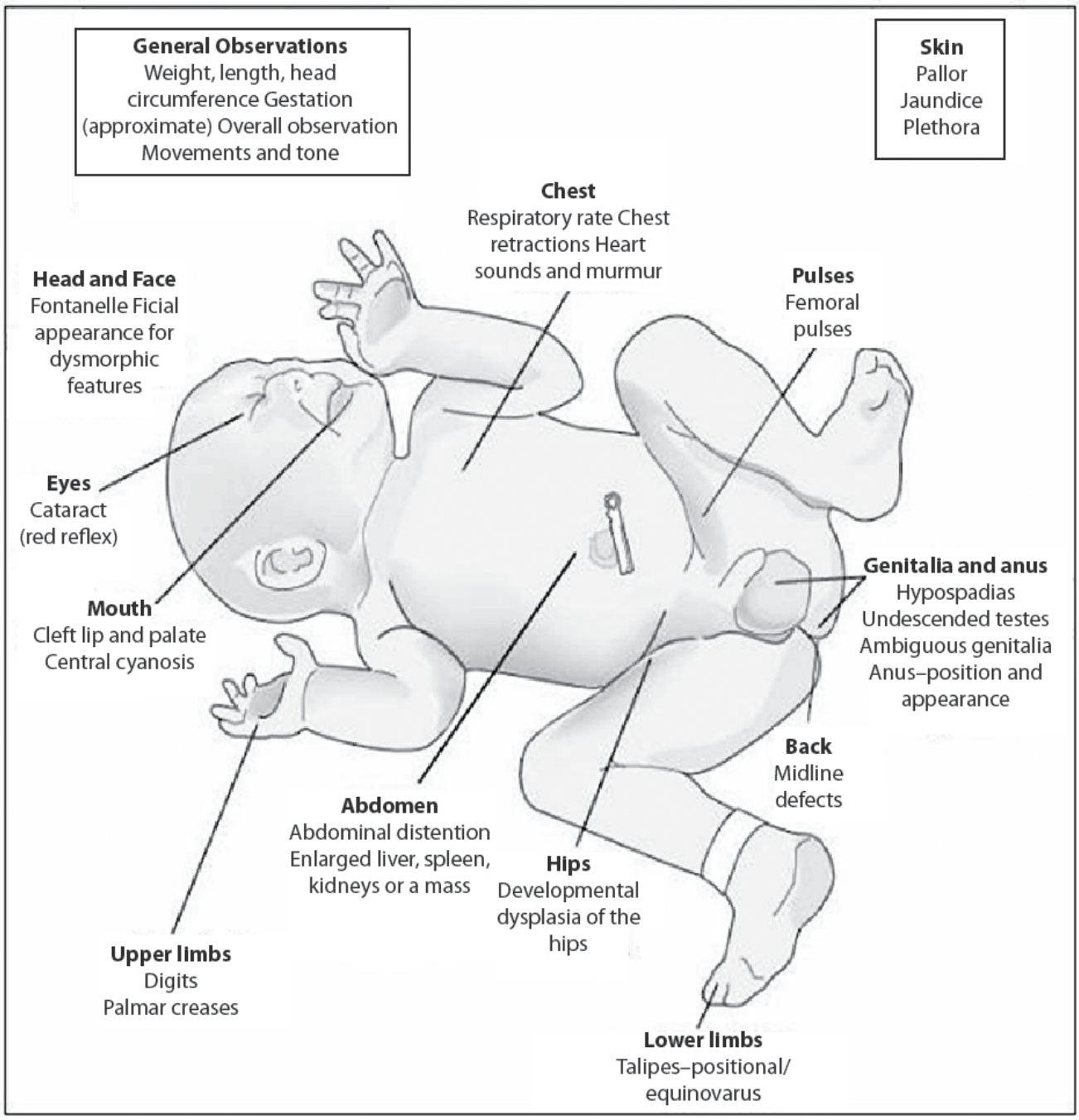
2.2 PROCESS FLOW



2.3 HEAD TO TOE EXAMINATION

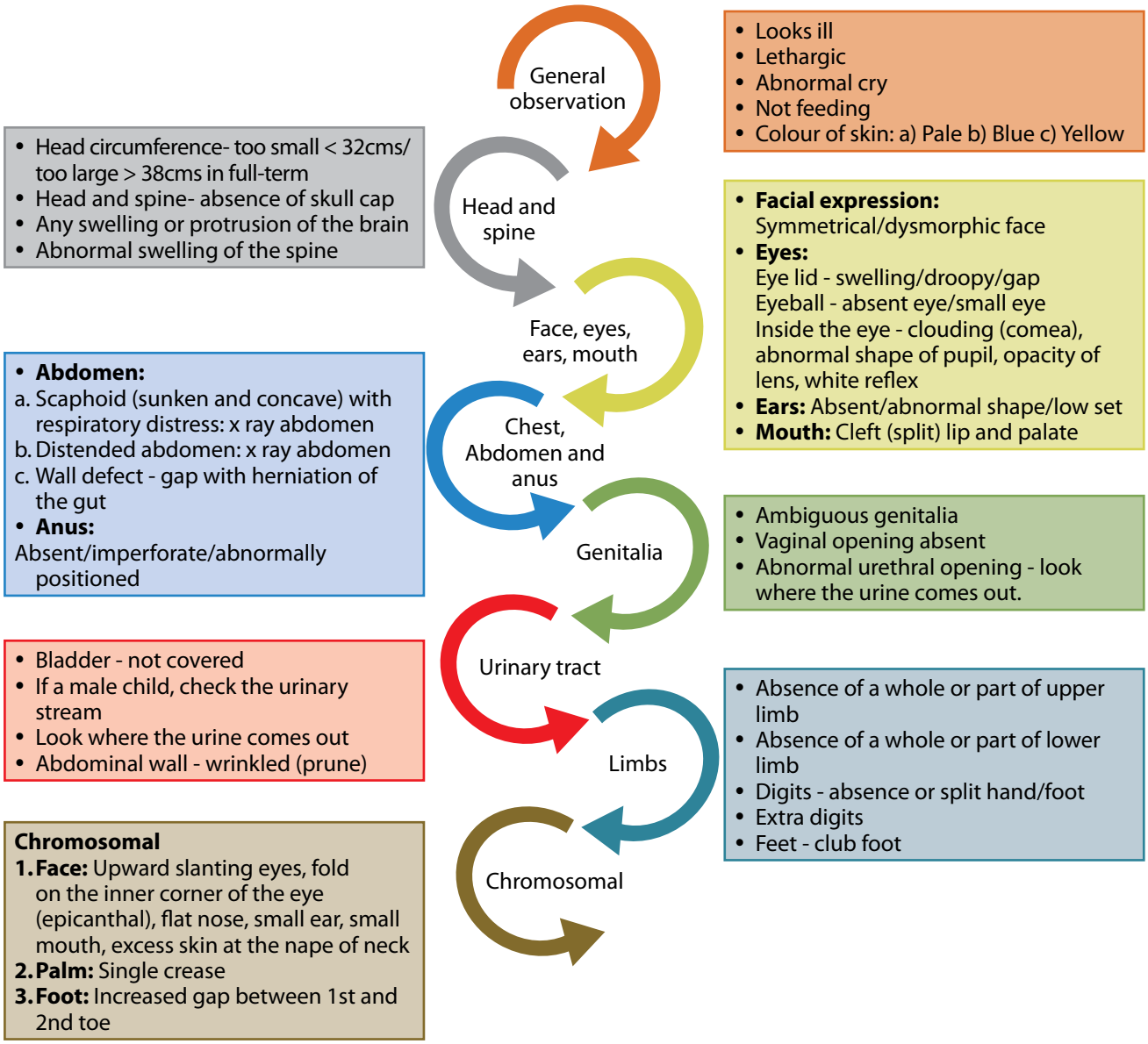
Check for Vital Signs (HRPT) before you preform the examination for Birth Defects. If the vital signs are abnormal, refer urgently		
Normal	Heart Rate (rates/min) Awake 100-180 Sleeping 80-160	Respiratory Rate (breaths/min): 30-60
Abnormal	Tachycardia: if more than 160 during sleeping	Tachypnea: if persistently more than 60 per minute
Physical Examination		
Aspect	**Clinical assessment**	**Indications for further examination and ☑ Urgent referral**
General Appearance	Examine the Newborn when quiet, alert, not hungry or crying State of alertness/ responsiveness to stimulus, Activity, spontaneous movement, predominant Posture	☑ *the child looks ill, or lethargic or has an abnormal cry* or abnormal movements
Head	• Shape and symmetry • Scalp swelling/deformity • Anterior and posterior fontanelle • Head circumference • Scalp lacerations/lesions	☑ Absence of cranial vault ☑ Herniation of the brain through a defect in the skull ☑ Closed fontanelles and fused sutures ☑ Enlarged, bulging or sunken fontanelle • Microcephaly/macrocephaly ☑ Hydrocephalous
Spine	• Spinal column • Scapulae and buttocks for symmetry • Skin over the spine	• Abnormal swelling of the spine • Non-intact bony spine • Abnormal curvature of spine • Tufts of hair or dimple along intact spine
Face	Facial expression Dysmorphic appearance Symmetry of structure	Asymmetry on crying ☑ Dysmorphic features
Eyes See pictorial tool for eye defects	1. Eyelid 2. Facial marks near the eye 3. Eyeball 4. Position in relation to the nasal bridge 5. Cornea 6. Pupil 7. Lens 8. Opacity with excessive tearing 9. Conjunctiva 10. White eye reflexes through a torch	1. Swelling, drooping or gap in the eye lid 2. Port wine stain or haemangioma 3. Abnormally small eye or absent eye 4. Upward slant/downward slant/epicanthic fold 5. ☑ Hazy, dull cornea, opacity 6. Pupils unequal, dilated or constricted or gap in the pupil 7. ☑ Congenital cataract 8. ☑ Congenital glaucoma 9. ☑ Purulent conjunctivitis
Ears	1. Family history or deafness 2. Position of ear 3. Shape of ear 4. Patency of the external auditory meatus	1. History positive 2. Abnormal placement of ear 3. Abnormal shape or absence
Mouth	1. Lips 2. Palate (hard/soft)	1. Cleft lip / cyanosis 2. Cleft palate
Chin, Neck and Clavicles	1. Chin 2. Neck 3. Clavicles (collar bone)	1. Small receding chin/micrognathia 2. Neck webbing/ Masses/swelling 3. Absence of clavicles

Heart, Chest	**1. Chest:** • Chest size, shape and symmetry • Number and position of nipples **2. Respiratory:** • Chest movement and effort with respiration • Respiratory rate • Breath sounds **3. Cardiac:** • Pulses –femoral • Position of apex beat • Pulse oximetry (optional)	☑ Signs of respiratory distress ☑ Apnoeic episodes ☑ Weak or absent pulses ☑ Positive pulse oximetry screen (if performed)
Abdomen and Anus	**1. Abdomen** • Shape and symmetry • Defect in the abdominal wall • Umbilicus including number of arteries • Any abdominal mass **2. Anus** • Position • Patency	☑ Abdominal swelling: intestinal obstruction ☑ Abdominal scaphoid with respiratory distress ☑ Defect in the abdominal wall: Gastroschisis/ exomphalos • Less than 3 umbilical vessels ☑ Abnormal abdominal mass • Abnormal position of anus ☑ Absence, imperforate anus ☑ No meconium passed within 24 hours
Genitalia	**1. Male genitalia** • Penis including foreskin • Testes (confirm present bilaterally and position of testes) including any discolouration • Scrotal size and colour • Other masses such as hydrocele **2. Female genitalia** • Clitoris • Labia • Hymen • Urethral opening	**1. Male genitalia** • Micropenis (stretched length less than 2.5 cm) • Bilateral undescended testes ☑ Testicular torsion ☑ Unequal scrotal size or scrotal dis-colouration **2. Female genitalia** • Absence of vaginal opening • Pseudomenses **3. Either male/female** • Inguinal hernia/swelling ☑ Ambiguous genitalia
Urinary tract	Bladder wall Has the Newborn passed urine? Urethral opening: look from where the urine comes out Check for urinary stream in a male child	☑ Bladder wall not intact- bladder exstrophy ☑ No urine passed within 24 hours ☑ Posterior urethral valve-disrupted flow • Hypospadias/epispadias
Limbs	**1. Upper Limbs** • Arm • Forearm • Hand, digits and palm **2. Lower Limbs** • Thigh • Leg • Foot and toes	• Absence of the whole or a part of the upper limb (arm/forearm, hand) • Extra digits/webbing of fingers • Single transverse crease • Absence of the whole or a part of the lower limb • Clubfoot
Hip	• Check symmetry of the legs • Skin folds over the buttocks • Risk factors for hip dysplasia: a. breech b. females c. family history	Hip dysplasia
Chromosomal	Look for any dysmorphic feature. Look at the face for upward slanting eyes, epicanthic fold, flat nose, small ears, small mouth, single palmar crease and increase gap between the first and second toe.	☑ Presence of chromosomal disorder

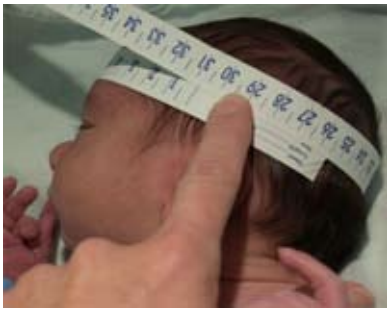


Before touching the baby, wash your hands

SEQUENCE OF EXAMINATION



MEASURING HEAD CIRCUMFERENCE



- Use a non-stretchable tape. Place it on the most prominent point at the back of the skull (the occiput) and just above the eyebrows (on the super-ciliary ridge).
- The measuring tape passes just above the eyebrows and around the prominent posterior aspect of the head.
- If the child has any protuberance on his or her forehead that makes it asymmetrical, put the tape over the most prominent part.
- Take the measurement in cm.

FLOW CHART: ROUTINE NEWBORN ASSESSMENT FOR BIRTH DEFECTS

Preparation

Essential steps

- Wash your hands before you touch the baby
- Seek parental consent for examination
- Encourage breastfeeding
- Encourage bonding

Timing

- Initial exam immediately after birth check the vitals. if present any, refer/ resuscitation
- Full and detailed assessment for Birth Defects prior to discharge
- Follow-up in high risk cases
- If unwell/premature – Refer if clinically indicated

Review history

- Maternal medical/obstetric/social
- Family history of Deafness, childhood blindness.
- Family history of congenital heart disease

Environment

Ensure:

- Warmth, lighting
- Correct identification
- Infection control precautions
- Privacy

Equipment

- Overhead warmer if required
- Stethoscope
- Pencil torch
- Tape measure, infant scales, growth charts, especially for head circumference
- Pulse oximetry (optional)
- Documentation
  - only if a Birth Defect is identified

Review discharge/refer criteria

- Stabilise before discharge or refer
- Observation for cry, colour, feeding, urine, stool

Discuss

- Place of referral
- Routine immunisation

Assessment areas

General appearance

↓

Head and Spine

↓

Face, Eyes, Ears

↓

Mouth and Lips

↓

Chest- respiration, Heart

↓

Abdomen, Anus

↓

Genitalia

↓

Urinary tract

↓

Upper limb & lower limb

↓

Chromosomal- down syndrome

↓

Discuss, document refer

Further examination ☑ urgent referral

Growth and appearance

- Dysmorphic features
- Excessive weight loss

☑ Bilious vomiting: X-ray abdomen

✓ Jaundice < 24 hours of age or on palms and soles

✓ Central cyanosis

Head and neck

✓ Enlarged/bulging fontanelle (hydrocephalus)

- Macro/microcephaly

✓ *Fused sutures*

- Facial palsy/asymmetry on crying
- Corneal opacity
- Congenital glaucoma
- Absent red eye reflex
- Pupils unequal/dilated/constricted
- Purulent conjunctivitis

Non-patent nares (bilateral nasal obstruction)

- Cleft lip/palate
- Unresponsive to noise
- Absent ear canal or microtia
- Small receding chin/micrognathia
- Neck masses, swelling, webbing

Upper limbs

- Limb hypotonia, contractures, palsy
- Palmar crease pattern

Chest

✓ Respiratory distress: X-ray chest

✓ Apnoeic episodes

✓ Weak or absent pulses

✓ Positive pulse oximetry

Abdomen and anus

✓ Gastrochisis/omphalocele

✓ Bilateral undescended testes

✓ Intestinal obstruction

✓ Imperforate anus

- Inguinal hernia
- < 3 umbilical vessels
- Signs of umbilical infection

Genitilia and urinary tract

✓ No urine/meconium in 24 hours

✓ Ambiguous genitalia

✓ Bladder exstrophy

✓ Posterior urethral valve

✓ Testicular torsion

- Hypospadias

Limbs

- Risk factors for hip dysplasia
- Contractures/hypotonia
- Fixed talipes
- Developmental hip dysplasia

Neurological

- Weak/irritable/absent cry
- Absent reflexes
- No response to consoling
- Inappropriate carer response to crying

Seizures

Altered state of consciousness

Chromosomal: Down syndrome

3. IMPLEMENTATION MODALITIES

3.1 THINGS TO ENSURE

1. RBSK Nodal Officer should ensure the availability of printed screening cum reporting format and monthly Birth Defect reporting format at all delivery points.
2. Ensure that the poster: 'examination of the Newborn from head to toe', is displayed at delivery points.
3. A one day training to all ANM, Staff Nurses and Doctors involved in the delivery of Newborn, using this handbook, to help them identify Visible Birth Defects, perform initial management and learn to fill in the relevant recording and reporting forms.
4. Ensure basic Newborn care to all Newborns regardless of having Birth Defect or not.

3.2 THINGS TO DO

1. Wash your hands before touching the baby.
2. Check for vital signs and if found abnormal, refer immediately.
3. Identify Birth Defect through physical examination-head to toe- with the aid of pictorial tool (poster: 'examination of the Newborn from head to toe').
4. Perform the basic steps of essential Newborn care.
5. Perform the initial management of Birth Defects.
6. Record the findings in the screening cum reporting format, RCH register along with the MCTS number.
7. Refer to a higher centre as specified by district and State administration.
8. Counsel parents using IEC materials.

Under RBSK, 9 common Birth Defects viz., Neural Tube Defects, Down syndrome, Cleft lip and Palate, Talipes (club foot), Developmental Dysplasia of the Hip, Congenital Cataract, Congenital Heart Diseases, Congenital Deafness and Retinopathy of Prematurity have been included, based on the following criteria:

a) The condition should be an important health problem amongst the local population.

b) There should be an accepted treatment for that condition.

However, the screening of Birth Defects at delivery points shall not be limited to the above mentioned conditions. **Any structural or functional anomaly has to be screened, documented and reported as per the recording and reporting formats.**

EXAMINATION OF THE NEWBORN FROM HEAD TO TOE FOR COMMON BIRTH DEFECTS

RBSK

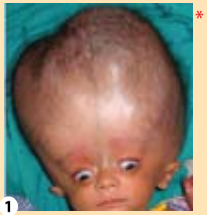
GENERAL OBSERVATION : If present, refer

• Looks ill • Lethargic • Abnormal cry • Not feeding • **Colour of skin:** a) Pale b) Blue c) Yellow

Wash your hands, before touching the baby

1 HEAD AND SPINE

1. Size too large > 38 cms (full term)
2. Size too small < 32 cms (full term)
3. Absence of skull cap
4. Swelling or protruding of the brain
5. Abnormal swelling of the spine


1 HYDROCEPHALOUS Q03 *


2 MICROCEPHALY Q02


3 ANENCEPHALY Q00

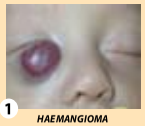

4 ENCEPHALOCELE Q01


5 SPINA BIFIDA WITH MENINGOMYELOCOELE Q05

2 EYES, EARS, MOUTH AND LIPS

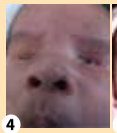
EYES

1. Eyelid – swelling
2. Eyelid – droopy
3. Gap in eyelid
4. Eyeball – absent
5. Eyeball – small
6. Inside the eye – corneal clouding
7. Inside the eye – opacity of lens/white reflex


1 HAEMANGIOMA D 18.01


2 PTOSIS Q 10.0


3 COLOBOMA OF EYELID Q10.3


4 ANOPHTHALMOS Q11.1 ONE EYE OR BOTH EYES

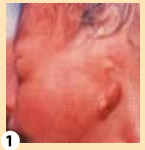

5 MICROTHALMOS Q11.2

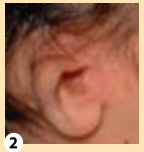

6 CONGENITAL CATARACT Q12.0 *

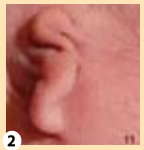

CONGENITAL GLAUCOMA Q15.0 *

EAR

1. Absent 2. Abnormal shape


1 ANOTIA Q 16.0


2 MICROTIA II Q 17.2


2 MICROTIA III Q 17.2

MOUTH

1. Cleft (split) lip 2. Cleft (split) palate 3. Cleft (split) lip and palate


1 CLEFT LIP Q 36

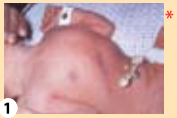

2 CLEFT PALATE Q 35


3 CLEFT LIP & PALATE Q37

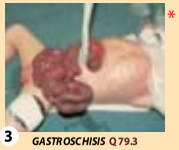
3 ABDOMEN AND ANUS

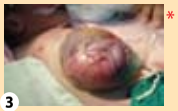
ABDOMEN

1. Scaphoid (sunken and concave) with respiratory distress: X-ray chest
2. Distended: X-ray abdomen
3. Wall defect- gap with herniation of the gut


1 DIAPHRAGMATIC HERNIA Q 79.0 *


2 INTESTINAL OBSTRUCTION Q 41-42 *


3 GASTROSCHISIS Q 79.3 *


3 OMPHALOCELE Q 79.2 *

ANUS

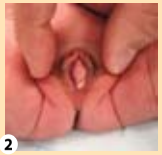
1. Absent/imperforate/ abnormally positioned


1 IMPERFORATE ANUS/ANORECTAL ATRESIA AND STENOSIS WITH OR WITHOUT FISTULA Q42.0-Q42.3 *

4 GENITALIA

1. Ambiguous genitalia
2. Vaginal opening absent
3. Urethral opening away from the tip of the penis – look where the urine comes out


1 AMBIGUOUS GENITALIA Q 56.4 *


2 VAGINAL AGENESIS Q 52.0


3 HYPOSPADIAS Q 54

5 URINARY TRACT

1. Bladder – not covered
2. Wrinkled abdominal wall
3. Urinary stream – check if male child


1 BLADDER EXSTROPHY Q 64.0-64.1 *


2 PRUNE BELLY Q 79.4


3 POSTERIOR URETHRAL VALVE Q 64.20 *

Distended bladder even after passing urine.

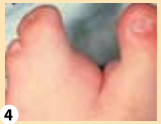
6 LIMBS (UPPER & LOWER)

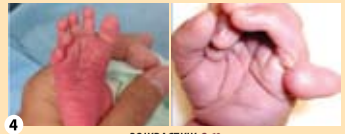
1. Absence of a whole or part of upper limb
2. Absence of a whole or part of lower limb
3. Fused digits
4. Absence of digits or split hand/foot
5. Extra digits
6. Club foot


1 LIMB REDUCTION DEFECT UPPER Q71


2 LIMB REDUCTION DEFECT LOWER Q72


3 SYNDACTYLY Q 70


4 ECTRODACTYLY Q 72.7


4 POLYDACTYLY Q 69


5 CLUB FOOT-TALIPES EQUINOVARUS Q 66.0

7 CHROMOSOMAL - DOWN SYNDROME

1. Face: Upward slanting eyes, fold on the inner corner of the eye (epicanthal), flat nose, small ear, small mouth, excess skin at the nape of neck
2. Palm: Single crease
3. Foot: Increased gap between 1st and 2nd toe


1 2 3 DOWN SYNDROME Q90

* Need uraent referral

→ *If anv of the above identified. record findinas in RCH reaister and RBSK birth defect recordina format alona with MCTS details.*

4. RECORDING AND REPORTING

- In order to capture Birth Defects at Delivery Points (DPs), a special recording form (Screening cum reporting form for Birth Defects) has been developed.
- ANMs/Staff nurse/Doctors at DPs are required to fill in the form at the time of birth of the child only if the Newborn has Birth Defect (s)*. Further, make corresponding entries in the labour room/delivery point register/RCH register.
- Maximum amount of information should be captured on the basis of history and clinical examination of the Newborn.
- The front side of the form consists of details to be filled. The back side of the form has a pictorial depiction of a child wherein the defect identified has to be ticked and the affected area is to be encircled/ticked. The variables are described in the annexure VIII.
- Individual recording formats are to be shared with the DEIC Manager who in turn will ensure evaluation, referral and follow up of these cases.
- A monthly reporting format indicating the total number of each category of Birth Defects is to be compiled at the block, district and state level and shared with the National RBSK unit of the MOHFW on or before 10th of every month.

BEFORE RECORDING THE FINDINGS:

- Know all the health conditions selected for identification of the Birth Defects along with the ICD 10 codes.
- Get familiarised with the case recording form and its fields.
- Develop the skills required for identification of visible Birth Defects.

* If a centre has got a delivery load of 25 cases per day (750 monthly or 9000 yearly), only 1-2 cases would present with a Birth Defect and hence the Staff Nurse would have to fill in only 1-2 forms per day.

RBSK: SCREENING CUM REPORTING FORM FOR BIRTH DEFECTS

FRONT SIDE:

1. Location of reporting	State	District	Block
2. Source of identification (please tick √) 01) Sub Centre 02) PHC 03) CHC/BPHC 04) Rural Hospital 05) Sub divisional Hospital 06) District Hospital 07) Medical College Hospital 08) Other Govt. Hospital 09) Tertiary centre 10) Any other Details of DP:			
3. Reporting Month: …/….		**4. Reporting Year:** …/…../…./…..	
5. Date of Identification (DD/MM/YY)	**6. Time of identification** O at birth O < 1 month O 1-12 months O 1-6 yrs. O above 6 years O Prenatal diagnosis O Spontaneous Abortion O Autopsy		
Delivery outcome details			
7. Date of Birth (DD/MM/YY)	8. Outcome of delivery: O Live Birth O IUD/Still birth, <20 wks.		
9. No. of Babies: O Single O Twins O Multiple	10. Birth weight in gms		
11. Sex O Male O Female O Ambiguous O Intersex	12. Gestational age (completed weeks):	13. Last menstrual period (DD/MM/YY)	
14. Birth asphyxia O Yes O No	15. Autopsy shows birth defect (if applicable): O Yes O No		
16. Status of induction/augmentation: O None O Oxytocin O Misoprostol			
17. Place of birth O Home O Institution	State	Dist	Block / Municipality
Identification details: MCTS/Unique ID/Aadhar (any one)			
18. a) MCTS: b) Unique ID* (computer generated): c) Mother's Aadhar no. (if available): d) Mobile no. of Mother:			
19. Child's name	20. Mother's name	21. Mother's age (in completed years)	
22. Father's age	23. Caste: O SC O ST O OBC O Others		
24. Permanent address: House No. Street name Area Post office District State			

Antenatal details (if available)							
25. Folic acid details (peri-conceptional)	O Yes O No	26. H/O serious maternal illness	O Yes O No	27. H/O radiation exposure (x-ray):	O Yes O No	28. H/O substance abuse	O Yes O No
29. Parental consanguinity Specify degree if known	O Yes O No	30. Assisted conception IVF/ART	O Yes O No	31. Immunisation history (rubella)	O Yes O No	32.Maternal drugs	O Yes O No
		If yes, describe				details, if available	
33. History of anomalies in previous pregnancies	O Yes O No	34. No. of previous abortion:		35. No. of previous still birth:			
If yes, describe							
Neonatal details							
36. Head circumference:		37. Birth defects: O Single O Multiple					

38. Visible Birth Defects (1. Head and spine 2. Face, eyes, ears, mouth and lips 3. Abdomen and anus 4. Genitalia 5. Urinary tract 6. Limbs (upper and lower)7. Chromosal disorders

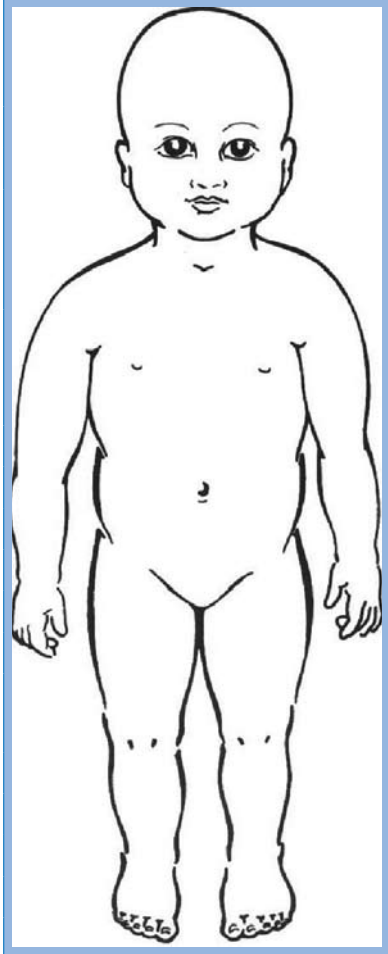
S.N	Type & Site	Description of the anomaly	Age at diagnosis	Code- ICD10	Confirmed or suspected
a.					
b.					

- This is the first page of the comprehensive Birth Defect recording format to be filled in by the Staff Nurse. Complete format is given in annexure VII.

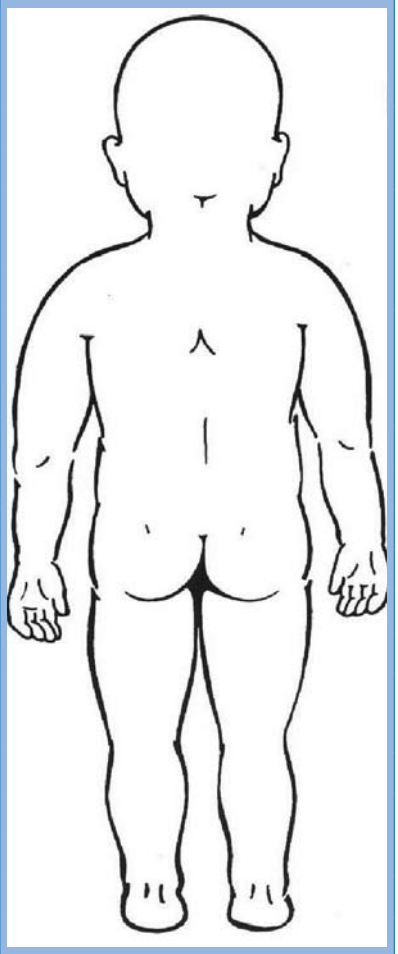
Section A of the format is to be filled in by the Staff Nurse and Section B by the Medical Officers/Paediatrician.

- For instructions to fill in this form, please refer to annexure VIII.

BIRTH DEFECTS ENLISTED FROM HEAD TO TOE



Body Part	Visible
Head & Spine	• Neural Tube Defect- Anencephaly, Encephalocele, Spina Bifida • Arhinencephaly/ Holoprosencephaly • Hydrocephalus • Microcephaly
Face	Asymmetrical/dysmorphic
Eye	
• Eyelid	Hemangioma, Ptosis (partial closure of the lid), Coloboma (gap)
• Eyeball	Anophthalmos, Microphthalmos
• Inside eye	Corneal clouding, Coloboma of Iris, Congenital cataract, Congenital glaucoma
Ear	Anotia, Microtia, Low set ears
Mouth and Lips	Cleft lip, Cleft palate, both
Abdomen and Anus	
• Scaphoid & sunken	Diaphragmatic hernia
• Distended	Congenital intestinal obstruction
• Herniation of gut	Gastroschisis, Omphalocele
• Anus	Imperforate anus, anorectal malformation
Urinary tract	
• Bladder	Bladder exstrophy
• Urinary stream	Posterior urethral valve
• Lower Abdomen	Prune belly
Genitalia	
Ambiguous	Indeterminate sex
Urethral opening	Hypospadias
Limb	Limb reduction Defects, Club foot, Polydactyly, Syndactyly, Oligodactyly
Chromosomal	Down Syndrome Others
Others	



Monthly reporting form: This is to be compiled at the block, district and state level and cumulative numbers are to be shared with the National RBSK unit of MOHFW

State	No. of Districts	No. of blocks	No. of DPs	Month/Year	Total BDs identified
S.N	**Name of Defect Identified**			**Grand Total:**	
A. Head and Spine: Q00 to Q07				**Total:**	
1	Anencephaly (NTD): Q00				
2	Myelomeningocele with Spina Bifida (NTD): Q05				
2	Encephalocele (NTD): Q01				
3	Hydrocephalus (NTD): Q03				
4	Arhinencephaly/Holoprosencephaly:Q04.1, Q04.2				
5	Microcephaly:Q02				
6	Others				
B. Eyes, Ears, Mouth and Lips					
B.1 Eye: Q10 to Q15				**Total:**	
7	Anophthalmos: Q11.1				
8	Microphthalmos: Q11.2				
9	Congenital cataract: Q12.0				
10	Congenital glaucoma: Q15.0				
11	Haemangioma: D 18.01				
12	Ptosis: Q 10.0				
13	Coloboma of eyelid: Q 10.3				
14	Others				
B.2 Ears: Q16 to Q17				**Total:**	
15	Anotia: Q16				
16	Microtia: Q17				
17	Others				
B.3 Oro facial: Q35 to Q37				**Total:**	
18	Cleft lip: Q36				
19	Cleft palate: Q35				
20	Cleft lip and palate: Q37				
21	Others				
C. Abdominal Walls and GI Defects				**Total:**	
22	Gastroschisis: Q79.3				
23	Omphalocoele: Q79.2				
24	Diaphramatic Hernia: Q 79.0				
25	Intestinal Obstruction: Q 41-42				
26	Imperforate anus/anorectal atresia and stenosis: Q42.0/Q42.3				
27	Others				
D. Genital Disorders: Q50 to Q56				**Total:**	
28	Hypospadias: Q54				
29	Epispadias: Q 64				
30	Vaginal Agenesis Q 52.0				
31	Ambiguous genitalia: Q56.4				
32	Others				
E. Urinary Tract Disorders				**Total:**	
33	Prune belly syndrome: Q79.4				
34	Bladder Exstrophy: Q64.1				
35	Epispadius: Q64.0				

36	Posterior urethral valve: Q64.20	
37	Congenital hydro nephrosis (USG):Q62.0	
38	Multicystic renal dysplasia (USG): Q61.40	
39	Bilateral renal agenesis (Potters Syndrome): Q60.1	
40	Others	
F. Limb Related Disorders: Q65 to Q79		**Total:**
41	Upper limb reduction defect: Q71	
42	Lower limb reduction defect: Q72	
43	Polydactyly: Q69	
44	Syndactyly: Q70	
45	Club foot: Q66.0	
46	Others	
G. Chromosomal Disorders: Q90 to Q99		
47	Down syndrome: Q90	
48	Other chromosomal disorders	
Any Other disorders (Please specify)		

5. COUNSELLING THE PARENTS

WHAT IS COUNSELLING?

- Counselling is a process of education, communication and support by which a counsellor helps the family to cope with difficult situations, enabling them to make important decisions and find realistic ways to solve their problems.
- Counselling, therefore, helps families to make their own informed decisions and supports their choices, rather than simply telling them what to do.

STAFF NURSE SHOULD BE ABLE TO COUNSEL THE PARENTS BY:

Providing the right information to make informed decisions, find realistic ways to solve their problems and remove myths and misconceptions.

- Birth Defect is not due to any fault of the parents including their lifestyle, but due to natural circumstances that was beyond the control of either of the parents and reassure the parents and the family that the cause of their child's Birth Defect was not due to any fault on their part.
- Importance of early and timely intervention: Birth Defects, if not addressed early in life, can lead to developmental delays and disabilities causing permanent damage to the brain. 90% of the brain development happens during the first two years of life through the right kind of inputs from all the sensory system such as vision, hearing, smell, touch, taste, movements of limbs etc. Damage to any of these sensory systems will affect the brain growth.
- Focus on parent's acceptance; but the immediate priority should be bonding, especially between mother and her child, feeding and warmth through proper clothing.
- Inform about existing intervention and available services. The possible interventions are:
  - Medical treatment
  - Surgical treatment
  - Neurodevelopmental therapy (NDT) and rehabilitation
  - Genetic counselling and psychosocial support
- Importance of feeding and maintaining warmth during transport of infants.
- Prevention of Birth Defects in subsequent pregnancies.

Refer to annexure VI for detailed information

* The advice should be realistic, taking into account the availability of local and national level resources and facilities. The information regarding the services, based on the mapped service list shared by the State, must be given to the parents.

6. REFERRAL

Appropriate referral: One of the main purposes of routine screening of the Newborn is to screen for Birth Defects and this may result in a referral for a minor or potentially major problem. An appropriate referral is defined as one in which:

a. the defect at birth is timely identified and referred to a centre where proper infrastructure and domain experts are available.

b. the child is at risk and requires further investigations or intervention.

c. the parents have a concern regarding the defect in their Newborn, which if missed, could be detrimental to the child's health in future.

Thus, the referral could be done for any of the above three situations.

Steps

i. **D**irectory: Procure the directory of service providers for appropriate management of Birth Defects from the RBSK Nodal Officer. Based on this list, the Staff Nurse should help the parents to seek appropriate referral within the District or the State.

ii. **D**estination: Mention the referral facility and the contact person in the referral card.

iii. **D**ocumentation: Make proper referral notes in the referral card.

iv. **D**EIC Manager: Inform the DEIC Manager about the referred case and mention his/her mobile number in the referral card.

v. **D**uring transport: Appropriate care to be provided for feeding and warmth, apart from respiration and circulation.

Before you transport please check the vitals of the Newborn:		
Airway	a) Check for Patency of the airway (Y/N)	
Breathing Normal respiratory rate is 30-60 breaths/minute	a) Check for respiratory rate (N/Ab) c) chest rise (N/Ab) e) Stridor (Y/N) g) Retractions (Y/N)	b) Airway entry (N/Ab) d) breath sounds (N/Ab) f) wheezing (Y/N) h) Grunting (Y/N)
Circulation Normal heart rate: Sleeping- 80 to160 beats/minute; awake- 110 to 180 beats/minute	a) Heart rate (N/Ab) c) Skin perfusion - CRT (N/Ab)	b) peripheral pulses (N/Ab) d) mottling (Y/N)

Disability: CNS perfusion	1. Alertness: Arousal, Irritable, Lethargic, Unarousable even with painful stimuli 2. Consolability: Cries cannot be consoled: Inconsolable 3. Cry: No cry, whimpering cry, cries to stimuli but normal pitch, High pitch continuous 4. Posture: abnormal posture: a)Opisthotonus, b) marked leg extension with strong arm flexion, c) jerky movements d) Hypotonic (all limbs hanging loosely) e) continuous fisting or thumb adduction 5. Sucking swallowing: absent or abnormal 6. Vision: persistent nystagmus, strabismus, roving eye movements, abnormal pupil size 7. Abnormal Primitive Reflexes: a) Moro) ATNR *Arousal is defined by the duration of eye opening and spontaneous movement of the face and extremities.* *Irritable: An infant is one who is agitated and cries with minimal stimulation and is unable to be soothed. Lethargic infants cannot maintain an alert state.*

7. INITIAL MANAGEMENT

7.1 CARE OF THE INFANT WITH NEURAL TUBE DEFECT (MYELOMENINGOCELE)

Key Points:

- Infants with Myelomeningocele have a high risk of developing latex allergies. It is therefore important to avoid any contact with products containing latex. Instead, silicon, portex or vinyl gloves should be used.
- 90 to 95% of infants with an open myelomenigocele will develop hydrocephalus.
- Daily monitoring of head circumference and fontanelle size by medical staff is required.
- Care must be taken to protect the exposed meninges in the spinal lesion until surgical closure can be performed.

DRESSING FOR OPEN NEURAL TUBE DEFECTS (NTD)

- Handle the infants with sterile, non – latex gloves and with sterile clothing and sheets.
- Cover the lesion with non-adhesive dressing like paraffin gauze followed by wet saline gauze.
- Place one end of a small orogastric tube (size 6 Fr) between the two layers of saline gauze and connect the other end to a syringe containing sterile saline solution.
- Place a ring of curlex around the lesion to prevent pressure on the sac.
- Cover the entire lesion with cling wrap and make a single wrap around the infant's chest or abdomen to keep in the place.
- Inject 0.5 cc saline every 2 hours to keep the dressing moist.
- Change the dressing once per day or when soiled/displaced.

See annexure IV for illustrations

7.2 CLEFT LIP AND PALATE

FEEDING GUIDELINES FOR CLEFT LIP AND PALATE BABIES

- All babies to be encouraged for breast feeding.
- Assist with breast milk expression if breastfeeding not possible.

See annexure V for illustrations on how to feed a cleft lip/palate infant

MANAGEMENT ACCORDING TO CLEFT TYPE

Unilateral Cleft lip (palate not affected) *Place the child in an upright, sitting position.* Watch for a pattern for sucking and swallowing. Listen for a swallow followed by breath.	1. Use the breast to help seal the cleft/s. 2. Mother can place her finger over the cleft to occlude air entry and create a seal. 3. Breastfeed with the cleft uppermost, use modified football hold postion or dancer hand position. 4. Breastfeeding outcomes for a unilateral cleft lip should be positive: the breast partly obscures the cleft and the mother can focus on the whole baby during feeds.
Cleft Palate-narrow cleft of the hard palate	1. Breastfeeding – if baby can take the nipple and some of the breast tissue into the mouth the breast may form a seal stopping air from entering from the nose and breastfeeding proceed naturally. 2. For infants with Cleft Palate, it may be useful to position the breast toward the "greater segment" i.e., the side of the palate that has the most intact bone. This may facilitate better compression and stop the nipple being pushed into the cleft site. 3. Supporting the infant's chin to stabilize the jaw during sucking and supporting the breast helps to keep the breast in the infant's mouth.
Cleft Palate: wide unilateral or wide bilateral cleft of the hard palate, with or without lip and soft palate involvement	1. Initiate breastfeeding and observe if the breast tissue is able to seal the cleft. 2. When breastfeeding is not initially achieved, give expressed breast milk either through a spoon or a paladai and if larger volumes are required use a Nasogastric tube. 3. Babies with clefts involving the lip and palate would benefit by receiving a pre-surgical orthodontic plate within 2 weeks of life.
Clefts of the soft palate, unilateral, bilateral or bifid uvula	1. Initiate breastfeeding. 2. Observe for slipping off the breast and need for frequent re-latching. 3. Signs of milk transferring: See the rhythm- squeeze, suck, swallow, breath. 4. Any milk spilling from the nostrils. 5. The baby should complete the feed within 30 minutes.

7.3 BOWEL OBSTRUCTION

To manage bowel obstruction:

- Place the infant in an incubator for close observation and temperature control.
- Nurse the infant in a supine position with the head elevated.
- Place an orogastric tube (8 - 10FG) on low pressure suction (or aspirate with a syringe every 60 minutes and leave it open for free drainage). The amount and type (e.g. bile-stained, faeculent) of fluid aspirated should be recorded.
- Place nil by mouth.
- Commence IV fluids. Give maintenance fluids plus ml for ml replacement of nasogastric aspirate with normal saline.
- Obtain abdominal x-rays (include supine and erect or decubitus view).
- Note that a relatively gasless abdomen is compatible with mid-gut volvulus.
- Consult a pediatric surgeon to arrange transfer to an appropriate surgical centre.
- It may be appropriate to commence antibiotics preferably after blood culture is taken.
- Obtain blood for: blood sugar, total and differential count, electrolytes and blood grouping.
- Be aware that these infants frequently have associated problems of acidosis and shock.

7.4 ABDOMINAL DEFECTS

Management involves:

- Wrap abdomen & exposed organs of baby in cling film using sterile latex-free gloves. Cling film does not need to be sterile.
- Support the exposed organs to prevent occlusion of blood vessels.
- If a length of bowel appears to have impaired blood supply or drainage i.e. looks purple or black in one position, try gentle manipulation of the bowel into other positions to see if the circulation can be improved. The bowel may need to be rotated on its pedicle to achieve a better circulation.
- Cotton wool covering or the use of moist packs is contraindicated. (Cotton wool adheres to the bowel wall, cannot be fully removed and causes peritoneal granulomas; moist packs rapidly become cold and lead to hypothermia).
- Pass size 8 NG tube, leave on free drainage and aspirate every 60 minutes.
- Record colour & volume of aspirate.
- Place nil by mouth.
- Large fluid loss occurs when organs are exposed and the neonate may require vigorous fluid replacement. Start IV infusion - give usual Day 1 fluids.
- Monitor blood pressure closely.
- Check blood glucose immediately and monitor closely.
- Monitor temperature frequently.

- Patients with a ruptured exomphalos sac or gastroschisis may have major problems with temperature control due to evaporative heat loss.
- Arrange neonatal transfer to tertiary surgical center.
- Give antibiotics: Penicillin, Gentamicin (preferably after blood culture).
- Take blood for total count & differential count, electrolytes, blood culture, group and hold for cross match of blood.

7.5 AMBIGUOUS GENITALIA

Diagnosing a Newborn with ambiguous genitalia should be treated with urgency and the doctor on call should be informed. Ambiguous genitalia can be associated with hypoglycaemia and/or severe electrolyte disturbance. In addition, being told that their Newborn has an unknown gender can cause significant distress for parents.

Action: 1) Ask the family: Any Drug ingestion during pregnancy, previous siblings dying in the Newborn period or with over-virilisation or precocious puberty. 2) Examine for signs of hypoglycaemia or dehydration. 3) Examine for the palpable gonads in the labioscrotal or inguinal regions, penile length (Normal 2.5-4.5cm in full term infant), Position of the urethral opening, Labioscrotal fold fusion. Genitalia pigmentation. Send for serum electrolytes and glucose and start the IV fluid.

7.6 POSTERIOR URETHRAL VALVE

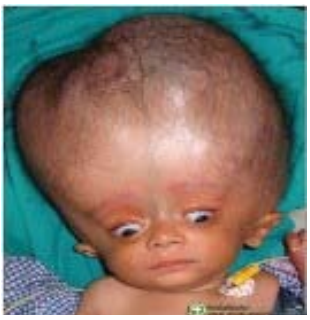
After delivery, bladder is catheterized with 6 Fr. feeding tube and antibiotics are administered. Infant is closely observed for hydration and electrolyte balance. Blood urea and creatinine are measured. Urinary volume, specific gravity, osmolality can be measured. Some may develop high output failure and will need appropriate fluid management. Babies may present in neonatal period with retention of urine, sepsis, failure to thrive, dehydration, acidosis, and pyuria or with vague symptoms and abdominal signs. These will need fluid resuscitation, catheterization, and administration of antibiotics, urine culture and sensitivity and monitoring of renal function. Priority in all patients with PUV is stabilization. Once the bladder is catheterized, it is no longer a surgical emergency.

ANNEXURE - I
ATLAS ON COMMON BIRTH DEFECTS WITH ICD 10 CODES

1. HEAD AND SPINE

Nervous system anomalies include a) Hydrocephaly, b) Microcephaly c) Neural tube defects d) other anomalies

Hydrocephalous Q03
(Hydro=water,cephalous=head)



- Dilatation of ventricular system, with or without enlargement of the skull.
- Identified by measuring head circumference more than 38 cms in a full term baby.

Microcephaly Q02
(Micro=small, cephaly= head)

- Reduction in the size of the brain.
- Identified by measuring the head circumference- less than 32 cms in a full term baby.

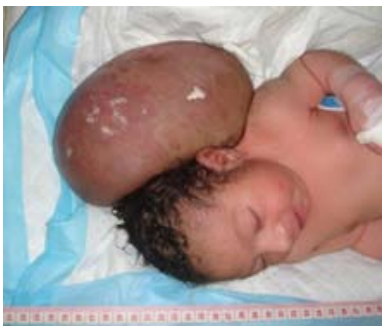
NEURAL TUBE DEFECTS: A) ANENCEPHALY B) ENCEPHALOCELE C) SPINA BIFIDA

- Neural Tube Defects (NTDs) are congenital defects of the brain and spinal cord.
- NTDs occur when the neural tube, the structure that develops into the brain and spinal cord, fails to close completely.
- NTDs may occur in the first few weeks, after conception.

Anencephaly Q00
(an=absent, encephaly = brain)

- Absence or deficiency of a major portion of the cranial vault, the covering skin and the brain tissue.
- Most of the brain and skull do not develop and babies are either stillborn or die shortly after birth.

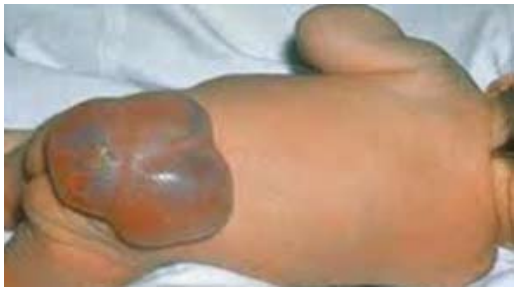
Encephalocele Q01
(Encephalo=brain,cele=herniation)



- Herniation of the brain and/or meninges through a defect in the skull.
- Presents as sac like protrusions of the brain covered with membranes through openings in the skull.

Spina Bifida Q05

(Spina= spine, bifida = split)



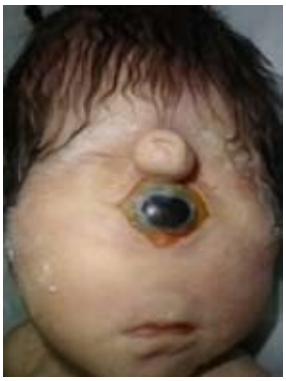
- Midline defect of the spine resulting in herniation (protrusion) of the spinal cord and/or meninges with or without a covering membrane.
- Closed defect is covered by normal-appearing skin.
- Commonly located in the lower or middle back or neck.

Arhinencephaly Q04.1

(A=absent, rhin= nose,encephaly=brain)

Holoprosencephaly Q04.2

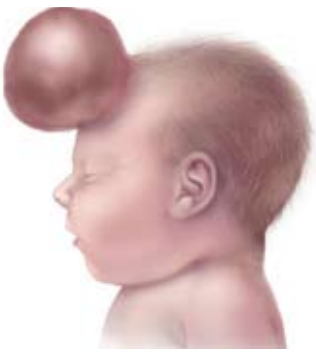
(Holo=whole, pros=fore or front, encephaly=brain)



- Arhinencephaly: Disorder in which the child has problem of smelling due to absence of part of brain associated with smell.
- Holoprosencephaly: Cephalic disorder in which the prosencephalon or the forebrain of the embryo fails to divide into two hemispheres.

➢ The picture is of a case of cyclopia, a form of holoprosencephaly, characterised by a single eye in the centre, absent nose and a proboscis (elongated protrusion) above the eyes.

Frontal encephalocele Q01.0



- Herniation of brain tissue, usually covered by meninges, through a defect in the frontal bone.

Nasofrontal encephalocele Q01.1

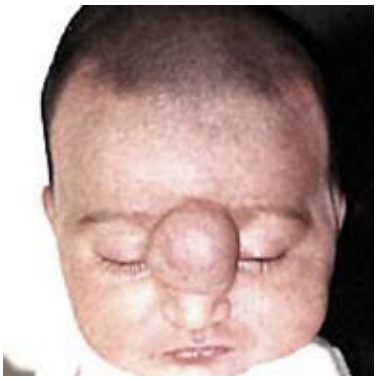
- Herniation of brain tissue, usually covered by meninges, through an opening between the frontal bone and the nasal bones.

** Photo Courtesy: WHO*

Occipital encephalocele Q01.2

- Herniation of brain tissue, usually covered by meninges, through an opening in the occipital bone.

Nasal encephalocele Q01.8



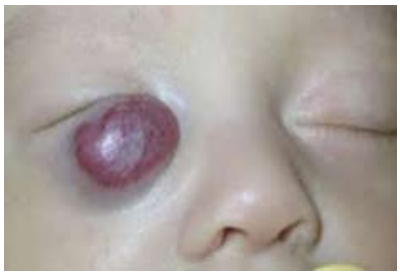
- Herniation of brain tissue through an opening in the nasal region.
- It is usually covered by meninges.

2. EYES, EARS, MOUTH AND LIPS

EYES

EYELID ABNORMALITIES

Haemangioma D18.01

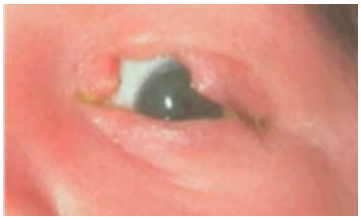


- Eyelid swelling.
- Presents as a red lesion or may be dark blue, if subcutaneous.
- May extend into the orbit and induce vision disorders like amblyopia.

Ptosis Q10.0

- Droopy eyelid.
- Partial closure of the eyes due to drooping of the upper eyelid.

Coloboma of eyelid Q10.3

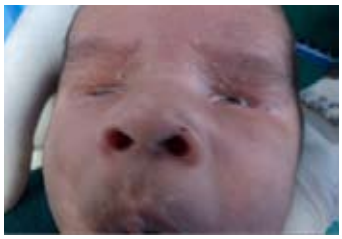


- Coloboma is a defect or gap in a structure of the eye like eyelid,iris, retina etc.
- The picture shows a gap or defect in the upper eyelid.

EYEBALL ABNORMALITIES

Anophthalmos of one or both eyes Q11.1

An =absent,opthalmos =eye



- Absence of eyeball or any ocular tissue.

Microphthalmos Q11.2

Micro =small, ophthalmos =eye

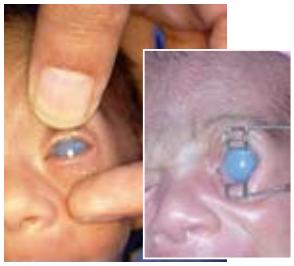
- Eyeball is abnormally small.

ABNORMALITIES INSIDE THE EYE

Congenital cataract Q12.0

- Opacity of the lens leading to a white reflex seen with or without a torch.

Congenital glaucoma Q15.0

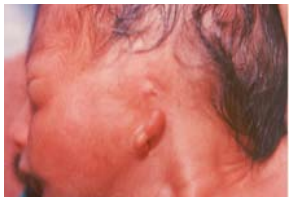


- Abnormally large eyes with corneal opacity with tearing eyes.

EARS

Anotia Q16.0

An =absent, Otia =ears



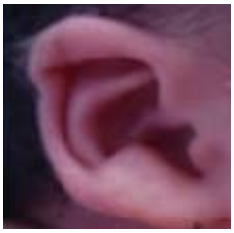
- Absence of ears.
- Picture shows a newborn with complete absence of external ears (auricles).

Microtia Q17.2

Micro=small, otia=ear

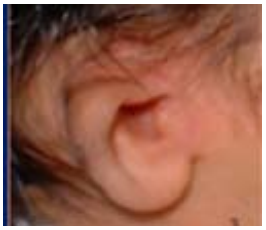
- Microtia is a congenital deformity of the outer ear where the ear does not fully develop during the first trimester of pregnancy.
- Microtia ears can vary in appearance (types I, II and III) but are usually smaller in size.

Microtia I



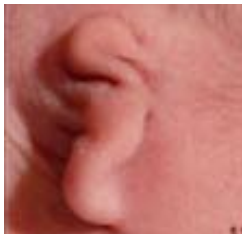
- Microtia I- The key features of the normal ear are present though they may have minor alterations in shape or form.

Microtia II



- Microtia II-Some of the features of the ear are missing, though usually there is still a lobule.

Microtia III



- Microtia III-This is the most common type of microtia, in which the only feature remaining is a small peanut-shaped remnant ear lobe.

MOUTH AND LIPS

Cleft lip Q36

- Physical split or separation of the two sides of the upper lip.

Cleft palate Q35

- Split or opening in the roof of the mouth connecting the nose.
- Usually associated with cleft lip but when alone may not be visible externally unless examined.

Cleft lip and palate Q37

- Bilateral cleft lip and palate giving a highly malformed appearance to the face.

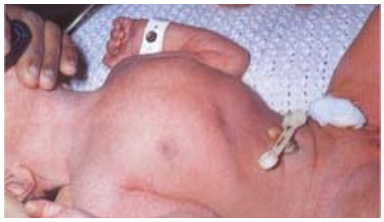
3. ABDOMEN AND ANUS

Inspection for

1. Scaphoid abdomen.
2. Distension of abdomen.
3. Defect in abdominal wall (gastroschisis).
4. Abnormalities of anus- absent, present but imperforate or abnormally located.
5. Any abnormality in umbilical vessels that normally have 2 arteries and one vein.
6. Hernias and Hydrocele- both umbilical and inguinal.

Palpation for any obvious swelling in the abdomen (*Perform when baby is sucking and relaxed and also ensure that your hands are warm).*

Congenital Diaphragmatic Hernia (CDH) Q.79.0

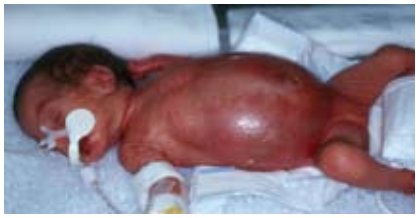


- Scaphoid (sunken and concave) or flat abdomen with respiratory distress seen in CDH occurs when the diaphragm does not form properly. (*The diaphragm is a thin sheet of muscle that separates the abdomen from the chest).*
- When there is an opening in the diaphragm, intestines that are normally in the abdomen, can get pushed (herniated) into the chest as shown in the figure.

Get an Urgent X-ray chest

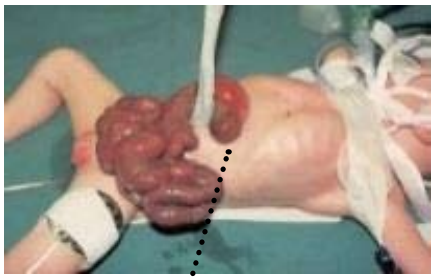
Congenital Intestinal Obstruction Q41-42

(High obstruction due to small bowel Q41; and Low obstruction due to large bowel is Q42)



- Abdomen is distended, may or may not be associated with vomiting.
- Usually stools are not passed within the first 48 hours, indicative of either a small bowel or large bowel obstruction.

Gastroschisis Q79.3

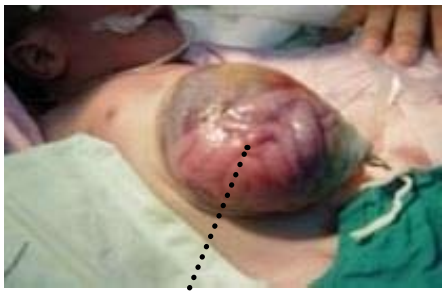


The intestinal loops are coming out from a defect that is to the right of the umbilicus with no covering

A birth defect of the abdominal (belly) wall. The baby's intestines stick outside of the baby's body, through a hole beside the Umbilicus with no sac.

- Not covered with a membrane.
- Defect is to the right of Umbilicus.
- Cord attached to abdominal wall.
- Immediate surgical consultation.

Omphalocele / Exomphalos Q79.2



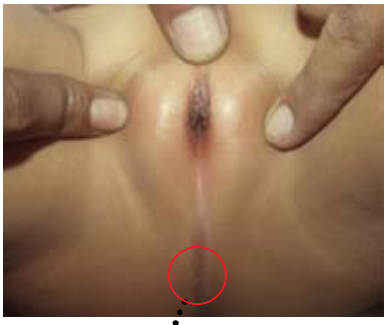
The intestinal loops are coming out from a defect at the umbilicus and covered by a sac

A birth defect of the abdominal (belly) wall at the umbilicus in which the intestines and occasionally other organs remain outside of the abdomen but covered in a sac.

- Covered with membrane unless it is ruptured.
- Cord attached at apex of the defect.

Immediate surgical consultation required.

Imperforate anus / anorectal atresia and stenosis with or without fistula Q42.0 -42.3



No anal opening seen

The anus is either not present or it is in the wrong place.

The variations could be:

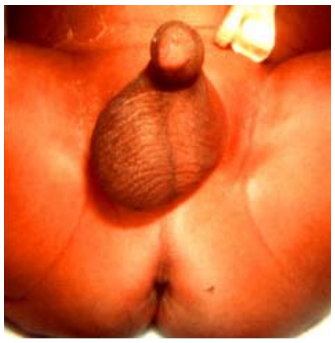
a. Anal opening is too narrow or in the wrong place.

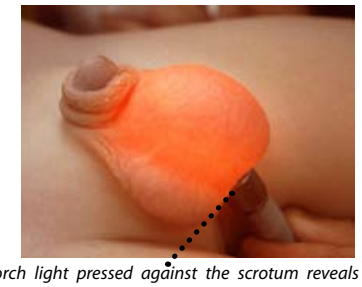
b. Membrane covers the anal opening.

c. Intestines are not connected to the anus.

d. An abnormal connection or fistula between the large intestines and urinary systems, allowing stool to pass through the urinary system and in girls may connect with the vagina.

Hydrocele (fluid filled sac or scrotum) P83.5

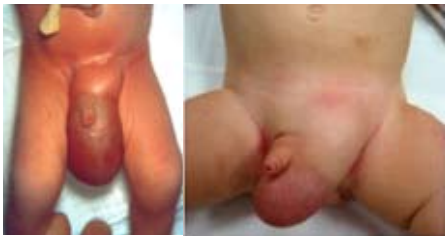




Torch light pressed against the scrotum reveals the watery nature of the fluid inside the swelling

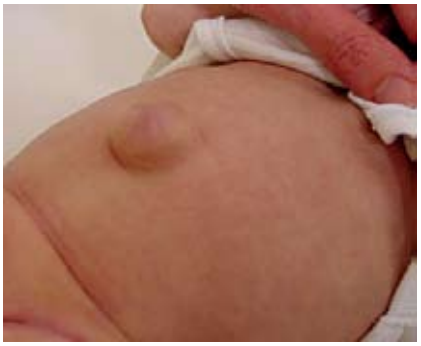
- It appears as a balloon like swelling of the scrotum or groin area due to trapping of fluid in the space between testes and scrotum ("Non-communicating hydrocele").
- In some cases, the swelling may extend up into the lower belly termed as "communicating hydrocele".

Inguinal Hernia K40



- An inguinal hernia appears as a bulge or visible lump in the groin on one or both sides due to a sac like projection of the abdominal cavity extending towards the scrotum (in boys) or labia (in girls).
- Sometimes a portion of the abdominal viscera may also migrate in the hernial sac.

Umbilical Hernia K42.9



- An umbilical hernia is an abnormal bulge that can be seen or felt at the umbilicus (belly button).
- This hernia develops when a portion of the lining of the abdomen, part of the intestine, and / or fluid from the abdomen, comes through the muscle of the abdominal wall.

4. GENITALIA

Male child

a. Penile size – A small penis (when less than 2 to 3 centimeters).

b. Urethral opening – In a male, the external opening of the urinary tract (external meatus) is normally located at the tip of the penis. Look for its position-away from the tip of penis or not.

Look from where the urine comes out.

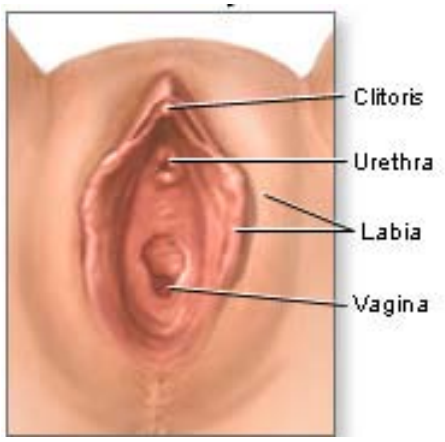
c. Palpation of Testes – If not palpable must determine if it is retractile, ectopic or cryptorchid. Unilateral retracted testicle may be brought down into scrotum.

Female child

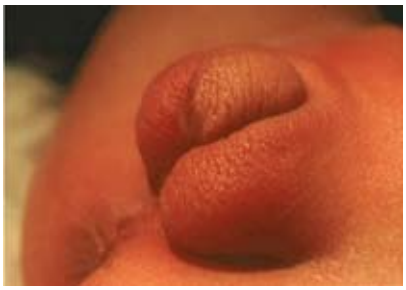
Female genitourinary examination check labia, clitoris, urethral opening (normally located between the clitoris and the vagina) and external vaginal opening.

Common normal variations:

1. Prominent labia minora and clitoris– Clitoris may have a relatively prominent appearance, especially if the labia are underdeveloped or the infant is premature.
2. Mild clitoromegaly.
3. Vaginal/hymenal skin tag and mild mucoid/whitish discharge.
4. Small amount of vaginal bleeding – Secondary to withdrawal of maternal hormone.



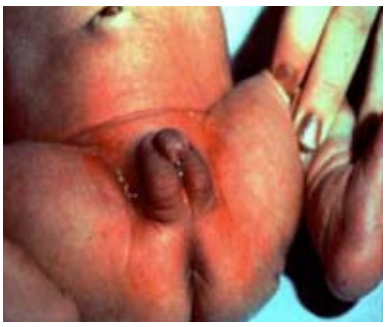
Ambiguous genitalia Q56.4



Ambiguous genitalia is a birth defect where the outer genitals do not have the typical appearance of either a boy or a girl.

a. Typically, ambiguous genitalia in true females i.e. genetic females (babies with X X chromosomes) has the following features:

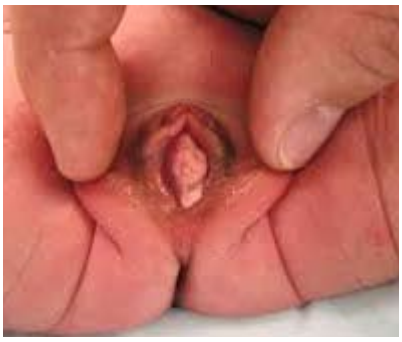
- An enlarged clitoris that looks like a small penis.
- The urethral opening (where urine comes out) can be anywhere along, above, or below the surface of the clitoris.
- The labia may be fused and look like a scrotum so the infant may be thought to be a male with undescended testicles. Sometimes a lump of tissue is felt within the fused labia, further making it look like a scrotum with testicles.



b. Typically, ambiguous genitalia in true males i.e. genetic females (babies with X Y chromosomes) has the following features:

- A small penis (less than 2 to 3 centimeters) that looks like an enlarged clitoris (the clitoris of a newborn female is normally somewhat enlarged at birth).
- The urethral opening may be anywhere along, above, or below the penis. It can be located as low as the perineum, further making the infant appear to be female.
- There may be a small scrotum that is separated and looks like labia.
- Undescended testicles commonly occur with ambiguous genitalia.

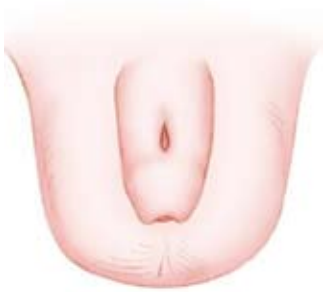
Vaginal agenesis Q52.0



- Vaginal agenesis is absence of vaginal opening, a congenital disorder of the female reproductive tract.

Epispadias Q64

(Epi means above)



Epispadius (in males):

- Abnormal opening of urethra on the upper side of the penis and may be at the level of neck of bladder rather than the tip.
- Short broad penis with upward curvature

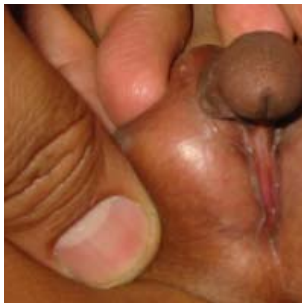
Epispadius (in females):

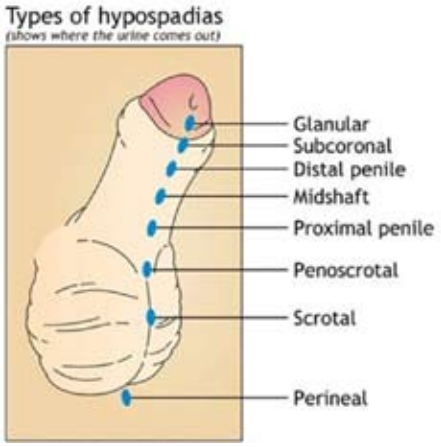
- abnormal opening between clitoris and labia, instead of between the clitoris and the vagina.
- Abormal clitoris, labia and wide pelvis (girls).

Epispadius is actually a milder expresssion of bladder exstrophy.

Hypospadias Q54

Hypo means under





Hypospadias (in males):

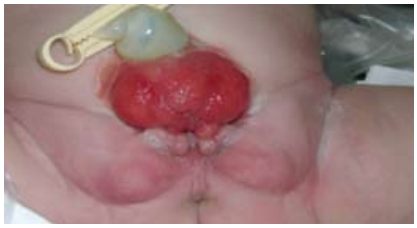
- Abnormal opening of urethra on the underside of penis rather than the tip.
- There is downward curvature of penis known as 'chordee' and
- A deficient prepucial skin on the underside.

The abnormal opening can be located anywhere between the perineum and the glans as shown in the figure.

Hypospadias (in females): the urethra opens into the cavity of the vagina.

5. URINARY TRACT

Bladder Exstrophy Q64.0-Q64.1



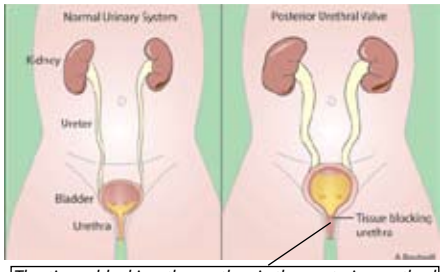
- Urinary bladder is malformed and exposed inside out, and protruded through the abdominal wall.
- Caused by closure defects involving the top front surface of the bladder, as well as the lower abdominal wall, skin, muscles and the pubic bone.

Prune Belly Q79.4

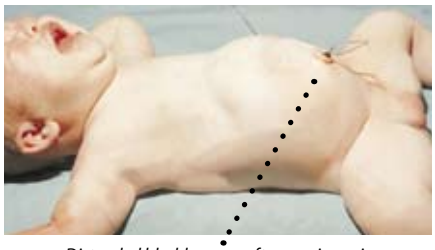
Condition characterized by: a) a lack of abdominal muscles, causing the skin on the abdominal area to wrinkle and appear "prune-like"; b) undescended testicles in males; and c) urinary tract malformations.

- The abdomen appears large and lax, the abdominal wall is wrinkled and shrivelled (prune like) with furrow like umbilicus.
- Skin folds may radiate from the navel or occur as transverse folds across the abdomen.

Posterior Urethral valve Q64.20



The tissue blocking the urethra is the posterior urethral valve causing backflow of urine leading to dilated ureters and enlarged kidneyswatery nature of the fluid inside the swelling

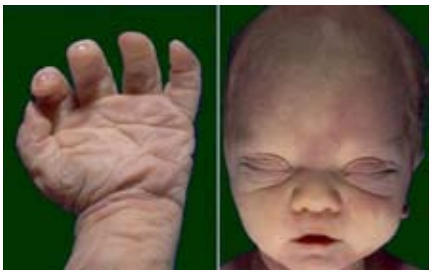


Distended bladder even after passing urine

A posterior urethral valve is an obstructing membrane in the posterior male urethra. It is the most common cause of bladder outlet obstruction in male newborns.

- A distended abdomen (bladder) even after passage of urine.
- The urinary flow would be drop by drop instead of a continuous stream Always check the urinary flow in a male child before discharge.

Potters syndrome Q60.6



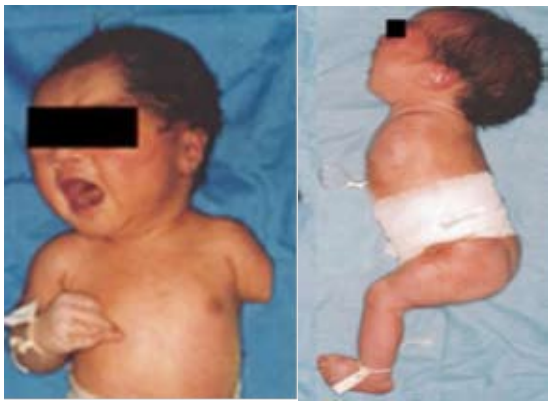
Refers to typical physical appearance especially that of face including widely separated eyes, and of limbs held in abnormal positions or contractures(wrinkled appearance) due to compression of the fetus as a result of markedly decreased amniotic fluid and associated pulmonary hypoplasia. It may be caused by bilateral renal agenesis.

6. LIMBS (UPPER AND LOWER)

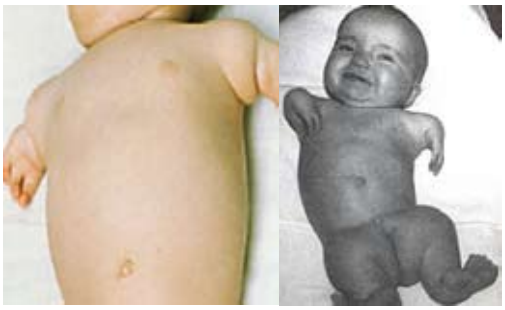
Congenital malformations and deformations of the musculoskeletal system or limb related disorders Q65-Q79

REDUCTION DEFECT OF UPPER LIMB

Congenital complete absence of upper limb Q71.0

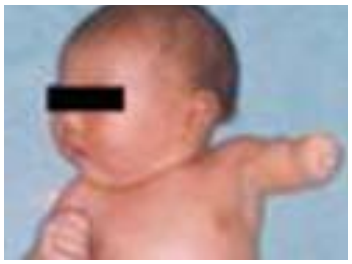


Congenital absence of arm and forearm Q71.1

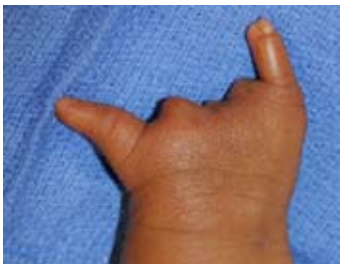


- Hands attached directly to the shoulder giving a seal fish appearance. The condition is referred to as phocomelia.

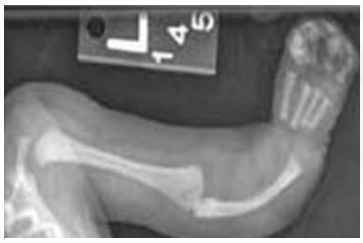
Absence of both forearm and hand Q71.2



Absence of hand and fingers Q71.3



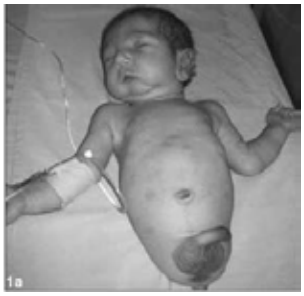
Longitudinal reduction defect of radius with either radial aplasia or hypoplasia Q71.4



Lobster claw hand Q71.6

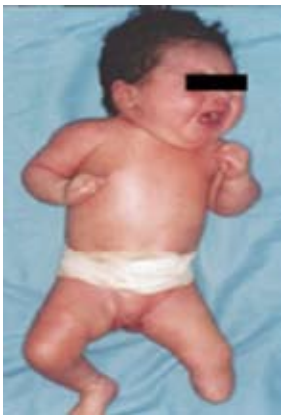
REDUCTION DEFECT OF LOWER LIMB

Congenital complete absence of lower limb(s) Q72.0

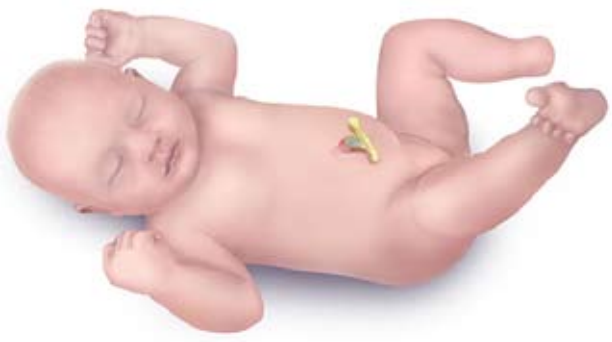


Congenital absence of thigh and lower leg with foot present Q72.1

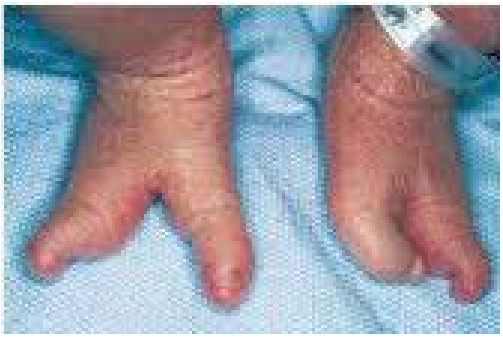
Congenital absence of both lower leg and foot Q72.2



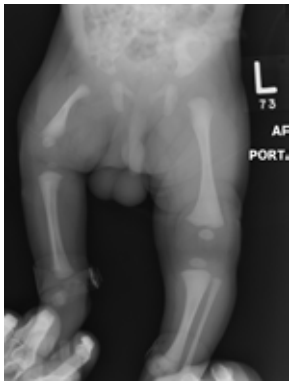
Congenital absence of foot and toe(s) Q72.3



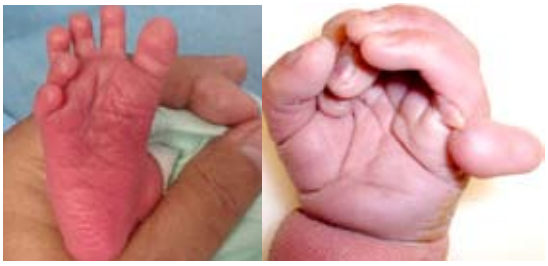
Split foot Q72.7



Congenital shortening of lower limb Q72.81

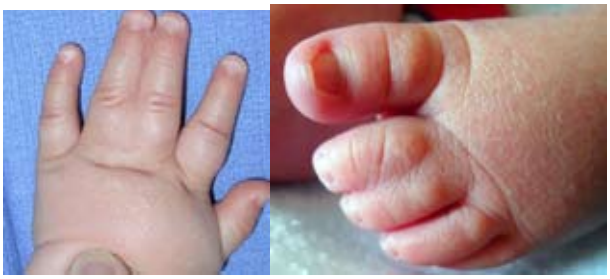


Polydactyly Q69



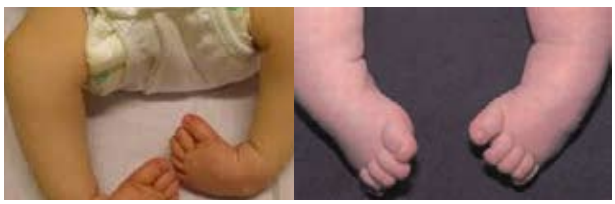
Extra or Accessory finger(s), Accessory thumb (s) or Accessory toe (s)

Syndactyly Q70



- Fused fingers and/or toes.
- Webbed figers and or/toes.

Club foot Q66.0

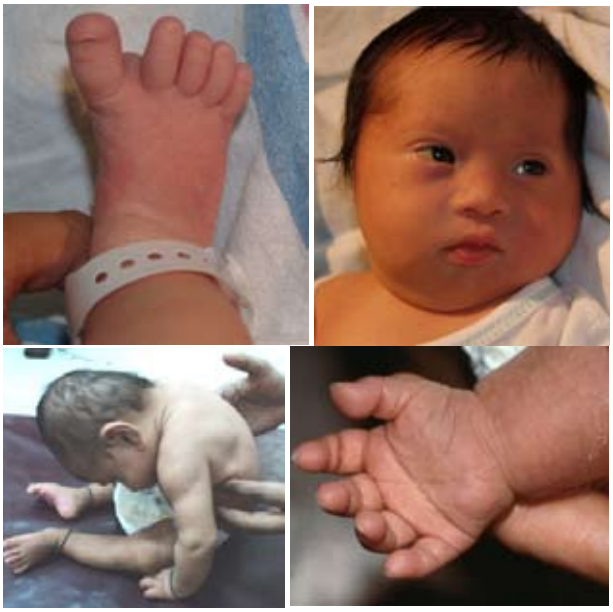


A congenital deformity of feet

- The foot (especially the heel) is usually smaller than normal.
- The foot may point downward.
- The front of the foot may be rotated toward the other foot.
- The foot may turn in, and in extreme cases, the bottom of the foot can point up.

7. CHROMOSOMAL – DOWN SYNDROME

Down Syndrome Q90

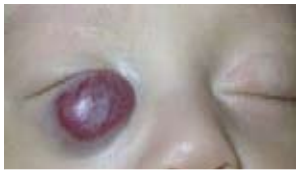


- Child appearing "floppy" i.e., head, body and hand hanging loosely.
- Head smaller than normal for same age group or abnormally shaped.
- Flat face with:
  a. Upward slanting eyes (may be normal in some parts of the country).
  b. Inner corner of the eyes rounded instead of pointed.
  c. Small and deformed ears.
  d. Small mouth.
  e. Flattened nose.
- Single deep crease across the centre of the palm.
- Fifth finger has one bending joint instead of two.
- Wide, short hands with short fingers.
- A big space between the first and second toe (sandal gap).
- Below average weight and length at birth.
- Enlarged appearing tongue in relationship to size of mouth.

ANNEXURE - II
PICTORIAL TOOL FOR IDENTIFICATION OF EYE DEFECTS

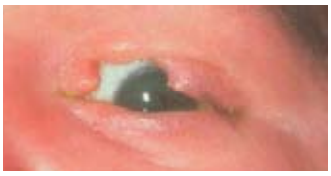
1. Check for Eyelid Abnormality

A. PARTIALLY CLOSED EYE LID, UNABLE TO KEEP IT OPEN DROOPY EYELID (PTOSIS)

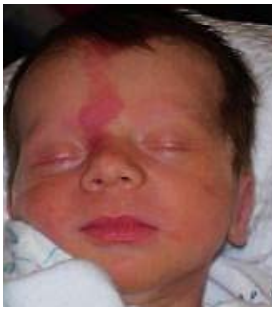


B. HEMANGIOMA OF EYE LID

C. EYE LIDS MAY BE TOO NARROW (BLEPHARO-PHIMOSIS)



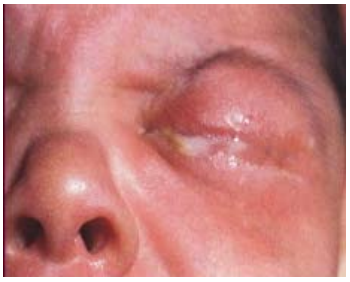
D. GAP IN EYE LID (COLOBOMA)

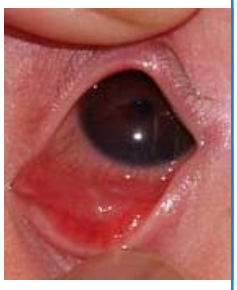


2. Check for Facial Marks: (Port Wine Stain or Capillary Hemangioma)

3. Abnormally Small Eye (Microphthalmos) or Absent Eye (Anophthalmos)

4. Look for Red Conjunctiva with Eye Discharge Ophthalmic Neonatorum





EYE DISCHARGE+ RED CONJUNCTIVA

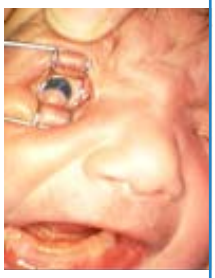
5. Coloboma:

A portion of the structure of eye is missing or a gap

KEY HOLE COLOBOMA OF BOTH EYES

6. Look for Blue Scelra and any growth on the Sclera

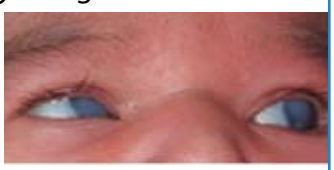
(Dermoid in Goldenhar Syndrome)



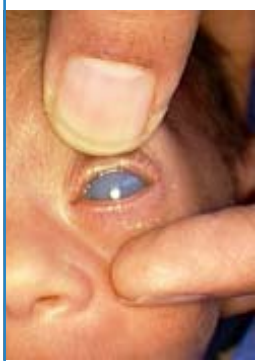
DERMOID

8. Pupil Area: Congenital Cataract Or Vitreous / Retinal Pathology

See for any white reflex by using a torch

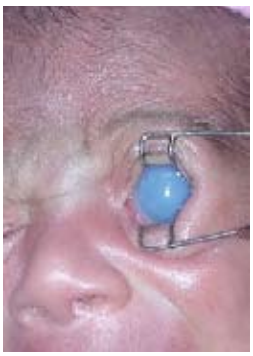


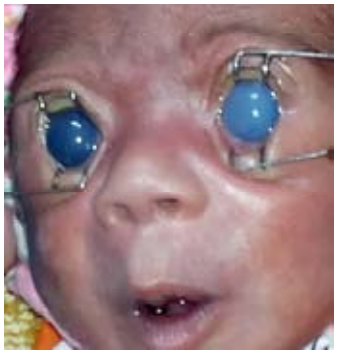
7. Corneal Opacity



CORNEAL CLOUDING

CONGENITAL CATARACT



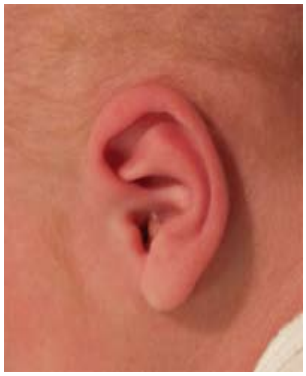


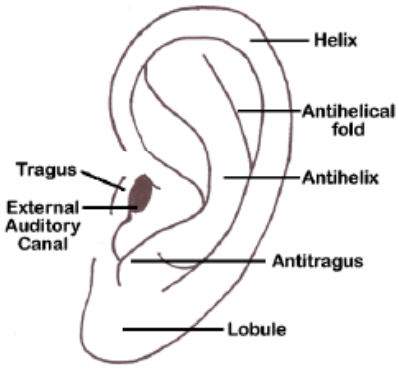
ABNORMALLY LARGE EYES WITH CORNEAL OPACITY: CONGENITAL GLAUCOMA IN NEW-BORN

ANNEXURE - III
PICTORIAL TOOL FOR EAR DEFECTS

ASSESS EAR POSITION RELATIVE TO THE FACE, FROM THE LATERAL VIEW

Ear shape and structure





NORMAL SHAPE, STRUCTURE AND PARTS OF EAR

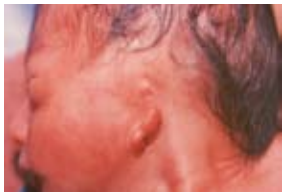
Abnormal shape structure and parts of ear

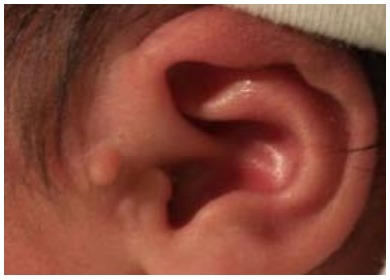
Abnormalities of external ear development is referred to as **Microtia.**

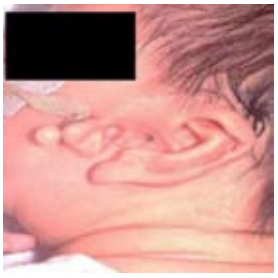
Abnormalities can be:

- Mild deviations
  - Ear size, shape, and location of the pinna and ear canal.
- Major malformations
  - Involving the external ear with only small nubbins of cartilage and an absent auditory meatus.

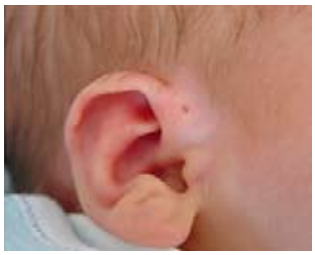
Complete absence of the pinna and ear canal is called **Anotia.**



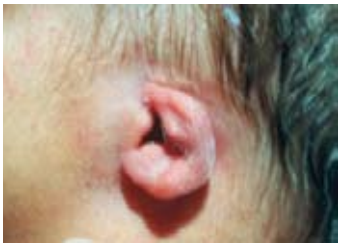




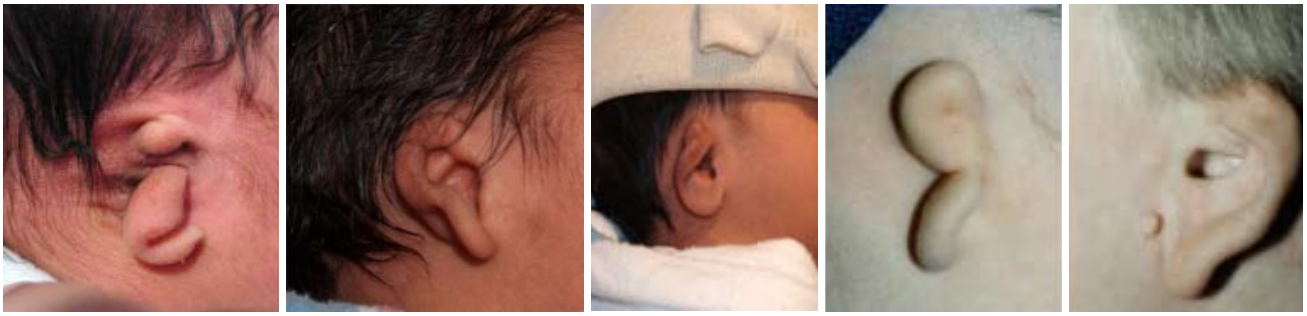
EAR TAG



PREAURICULAR EAR PIT



HYPOPLASTIC & LOW SET EAR



MICROTIA VARIANTS

ANNEXURE - IV
INITIAL MANAGEMENT OF NEURAL TUBE DEFECTS

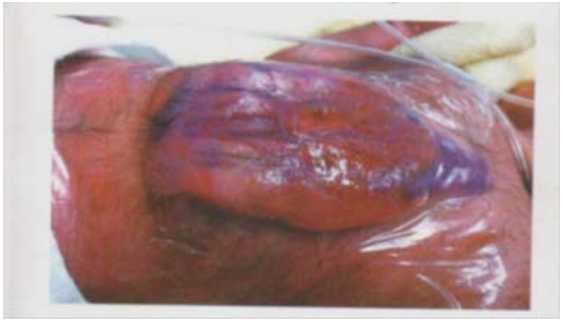
STEP - 1

Make a ring with cotton gauze of size, slightly larger than the lesion. Cover the ring with cling wrap to make it water resistant.

Cling Film Wrap

STEP 2

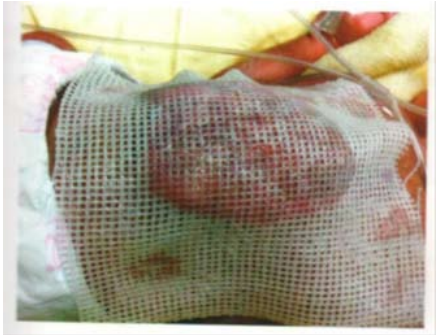
Place Tegaderm on the skin surrounding the lesion.



Tegaderm on the skin surrounding the lesion

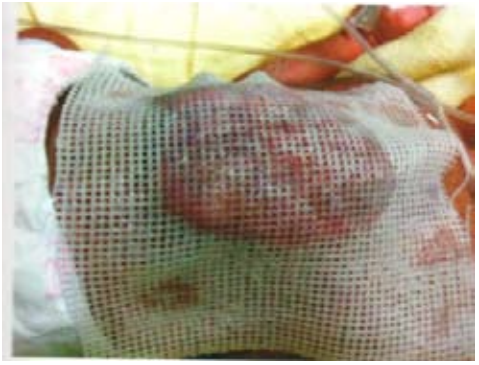
STEP 3

Cover paraffin gauze over the lesion.



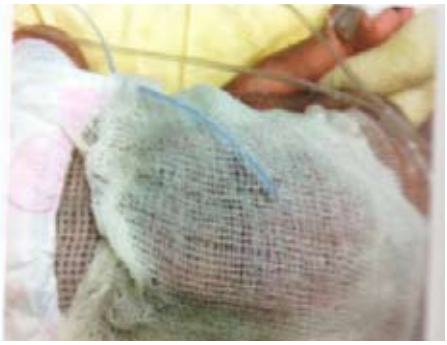
STEP 4

Place sterile saline soaked gauze over the paraffin gauze.



STEP 5

Insert the smallest OG tube between the two layers of gauze. Connect the other end to a syringe containing sterile saline.

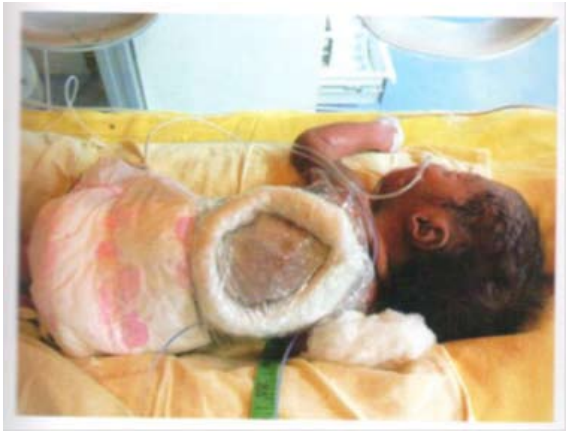


STEP 6

Place the ring around the dressing to hold it in place and to support the lesion.

STEP 7

Nurse the infant prone or side lying only. Inject 0.5 ml saline via the OG tube every 2 hourly to keep it moist.



Protected from soiling with a plastic flap. Care should be taken to prevent contamination as potential for infection is high.

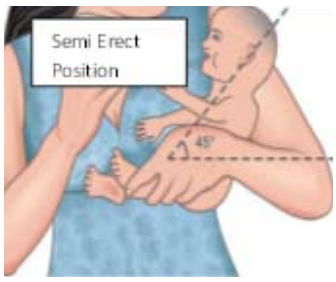
Minimal tape should be applied to the skin due to sensitivity to tapes and to prevent dermal stripping.

Counsel the mother of peri-conceptional intake of folic acid.

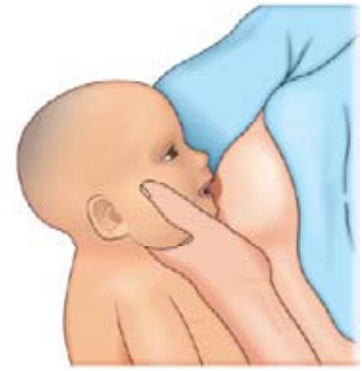
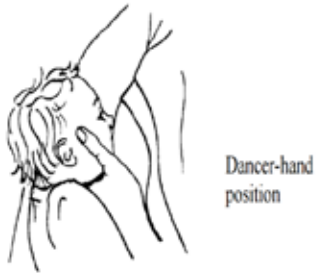
Courtesy: AIIMS Protocols in Neonatology

ANNEXURE - V
FEEDING A BABY WITH CLEFT LIP AND/OR PALATE

Babies with a cleft lip can usually breastfeed. Placing the baby in an upright position with the cleft covered by the mother's finger or breast tissue can aid attachment and reduce air intake. The secret is to create a seal where no air passes through. It just takes experimenting with a few positions and some are shown below. These positions help the milk from leaking from the nose and the dancer hold helps support the baby's chin.

<table>
<tr>
<td>  </td>
<td> 
</td>
<td>Place the child in an upright, sitting position. Watch for a pattern for sucking and swallowing.

Listen for a swallow followed by breath. The baby should complete the feed within maximum of 30 minutes.</td>
</tr>
<tr>
<td> 
Modified Foot ball hold postion</td>
<td> 

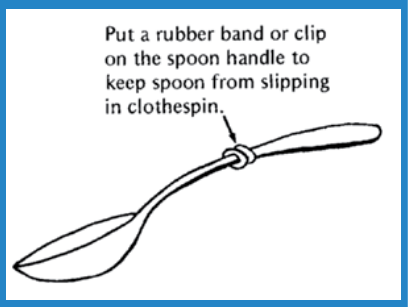
If the baby tends to choke and leak milk from the nose, the modified football hold may help.</td>
<td> 
While mother sitting on bed, position the infant in semi upright position (sitting position) with infants legs on the side of mother. The child's bottom should be on the bed and a pillow at breast level with pillow supporting the infants back while in sitting position. Mother can support the back and base of head with her arm. Mother may need to support her breast with the C hold: thumb on top and 4 fingers underneath the breast but away from areola.</td>
</tr>
<tr>
<td> 
Dancer Hand position</td>
<td> 

If baby has a non-rhythmic suck, try using a rocking chair.</td>
<td>This position can be used to support the infant's chin. Slide the hand under the breast forward i.e. supporting the breast with three fingers, placing underneath the breast and using the thumb and the forefinger form a U shape and support the baby's chin. This would help the baby to press the nipple and areola between the gums. Breastfeeding should be in calm surroundings with few distractions.</td>
</tr>
</table>

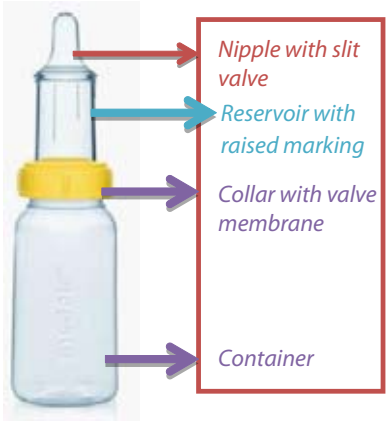
Feeding a baby with Cleft Lip and/or Palate

Spoon feeding with Express fresh breast milk of mother.

Tip of spoon should be soft and rounded with a long handle.



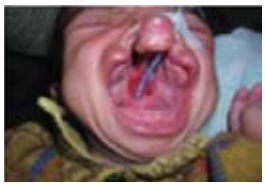
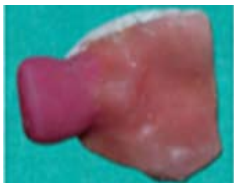
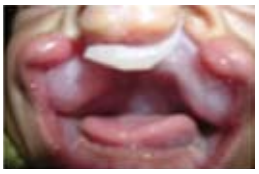
Paladai feeding: care to be taken to avoid pouring large amounts of milk in infants mouth.



Expressed milk being fed through **Haberman Feeder**. This is designed for babies with feeding problems. Special nipple not requiring suction with slit valve that controls the flow.

Mead Johnson cleft palate bottle for feeding expressed breast milk & is a squeezable bottle which can be squeezed rhytmically.

Feeding Obturator: feeding obturator is a prosthetic aid that is designated to close the cleft and restore the sepration between oral and nasal cavities. It creates a rigid platform and prevents the tongue from entering the defect and reduces choking and nasal regurgittation.

		
Bilateral Cleft Palate	Obturator	After Application of Obturator

ANNEXURE - VI
COUNSELLING PARENTS ON BIRTH DEFECTS

Who is at Risk for Birth Defects?

All pregnant women have some risk of delivering a child who suffers from a birth defect. Risk increases under any of the following conditions:

- Family history of birth defects or other genetic disorders.
- Drug use, alcohol consumption, or smoking during pregnancy.
- Advanced maternal age of 35 years or older.
- Inadequate prenatal care.
- Untreated infections or viruses, including sexually transmitted infections.

Preventing Birth Defects

Ensure that the pregnant woman:

- Does regular Antenatal checkup at least three times during pregnancy.
- Stays happy and stress free and rests at regular intervals.
- Folic acid intake should start soon after the marriage and continue during pregnancy. Also eat folic acid rich food like green leafy vegetables, liver and pulses.
- Goes for at least 4 antenatal check-ups.
- Do not smoke or drink alcohol.
- Minimize unnecessary medicines except those prescribed by ANM or medical officer.
- Maintain good hygiene by adopting safe sex practices and personal hygiene to prevent infection during pregnancy.
- Marriage among close relatives must be avoided, especially in families with a history of Birth Defects.
- Family members to maintain positive environment at home by avoiding any maternal stress and domestic violence.
- Immunization against rubella at least one month prior to pregnancy or during adolescence
- Always use iodized salt.
- Avoid taking any medications without consulting the doctor.
- Screening for diabetes during antenatal period and keep diabetes under control.
- Prevent infections by washing hands; cooking meat until it is well done, staying away from people who have infections; avoid cats during pregnancy.

- Always have safe and protected sexual activities.
- Be active and maintain a healthy weight.
- Avoid obesity and treat obesity before getting pregnant.
- Avoid exposure to hazardous environmental substance, heavy metals, pesticides and radiation exposures including X-rays.
- Consulting doctor for any family history of Birth Defects or genetic disease.
- Test for blood group and Rh compatibility.
- Timely referral of high risk cases of pregnancies including screening for advanced maternal age.

Neural Tube Defects

Neural tube defect:

Key messages to the parents:

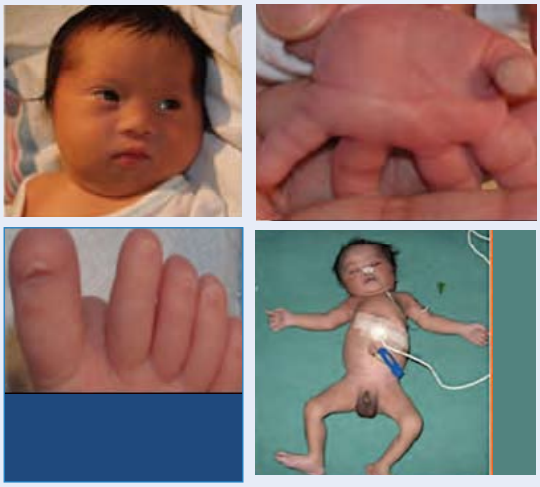
All pregnant women have some risk of delivering a child who suffers from a birth defect. Risk increases under any of the following conditions:

- Advise pregnant women to eat foods rich in folic acid like green leafy vegetables, pulses to prevent neural tube defects.
- Strictly avoid smoking/alcohol at any time and in any amount during pregnancy.

Down Syndrome

Key messages to the parents:

- With early intervention i.e., within the first three months, children with Down syndrome can lead a near independent life.
- Examination of eye, teeth, hearing and thyroid of the child should be done regularly.
- Special care of the child's neck and spine (avoid sudden movement to the neck)
- The child's speech can be improved if the parents are supported by a speech therapist.



Cleft lift and palate

Key messages to the parents

- Cleft lift and palate can be corrected by performing timely surgeries.
- By 2-3 months of birth, surgery is performed to close the cleft lip; cleft lip could be corrected before the child speaks.
- Cleft palate surgery should be performed between 12 to 18 months of age.

Club Foot

Key messages to the parents

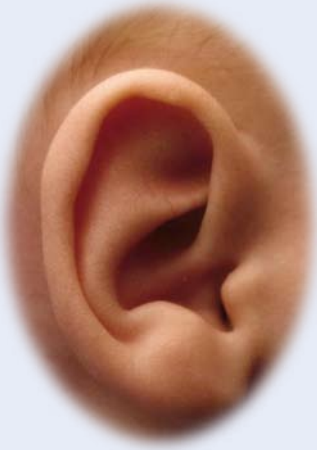
- Early detection and correction is important for child's development.
- If not treated at an early stage, this deformity can lead to lifetime disability.
- The mother should not try to correct the foot by pushing the sole towards the floor.
- Never pronate (rotate) and never put pressure on the heel
- Never use force while massage.
- Treatment to start within 7-10 days; requires multiple plaster 4-6 and changing weekly.

Congenital Cataract

Key messages to the parents

- This needs immediate referral, otherwise the child would become permanently blind
- If not corrected, the blindness would also affect the learning ability of the child

Congenital Deafness



Key messages to the parents

- Take the child for a hearing test at the District Hospital.
- Early management of hearing loss is important as it affects speech development.
- Reduce exposure to loud noise.
- Hearing devices, such as hearing aids and speech therapy can help in hearing.
- Every child has the ability to speak. Hence if a child's deafness could be confirmed by the age of 9 months and hearing aids could be provided before 2 years, the child could be prevented from becoming dumb.

ANNEXURE - VII

RBSK: SCREENING CUM REPORTING FORM FOR ALL BIRTH DEFECTS

FRONT SIDE

1. Location of reporting	State	District	Block
2. Source of identification (please tick √) 01) Sub Centre 02) PHC 03) CHC/BPHC 04) Rural Hospital 05) Sub divisional Hospital 06) District Hospital 07) Medical College Hospital 08) Other Govt. Hospital 09) Tertiary centre 10) Any other Details of DP:			
3. Reporting Month: …/….		**4. Reporting Year:** …/…../…./…..	
5. Date of Identification (DD/MM/YY)	**6. Time of identification** O at birth O < 1 month O 1-12 months O 1-6 yrs. O above 6 years O Prenatal diagnosis O Spontaneous Abortion O Autopsy		
Delivery outcome details			
7. Date of Birth (DD/MM/YY)	8. Outcome of delivery: O Live Birth O IUD/Still birth, <20 wks.		
9. No. of Babies: O Single O Twins O Multiple	10. Birth weight in gms		
11. Sex O Male O Female O Ambiguous O Intersex	12. Gestational age (completed weeks):	13. Last menstrual period (DD/MM/YY)	
14. Birth asphyxia O Yes O No	15. Autopsy shows birth defect (if applicable): O Yes O No		
16. Status of induction/augmentation: O None O Oxytocin O Misoprostol			
17. Place of birth O Home O Institution	State / Dist	Block	Municipality
Identification details: MCTS/Unique ID/Aadhar (any one)			
18. a) MCTS: b) Unique ID* (computer generated): c) Mother's Aadhar no. (if available): d) Mobile no. of Mother:			
19. Child's name	20. Mother's name	21. Mother's age (in completed years)	
22. Father's age	23. Caste: O SC O ST O OBC O Others		
24. Permanent address: House No. Street name Area Post office District State			

Antenatal details (if available)							
25. Folic acid details (peri-conceptional)	O Yes O No	26. H/O serious maternal illness	O Yes O No	27. H/O radiation exposure (x-ray):	O Yes O No	28. H/O substance abuse	O Yes O No
29. Parental consanguinity Specify degree if known	O Yes O No	30. Assisted conception IVF/ART	O Yes O No	31. Immunisation history (rubella)	O Yes O No	32. Maternal drugs	O Yes O No
		If yes, describe				details, if available	
33. History of anomalies in previous pregnancies	O Yes O No	34. No. of previous abortion:		35. No. of previous still birth:			
If yes, describe							

Neonatal details		
36. Head circumference:	37. Birth defects: O Single O Multiple	

38. Visible Birth Defects (Nervous system, Eye, Ear, face & neck, CHD, Respiratory, cleft lip & palage, digestive, genital, urinary, musculo skeletal, chromosomal, syndromes, skin, limbs)

S.N	Type & Site	Description of the anomaly	Age at diagnosis	Code- ICD10	Confirmed or suspected
a.					
b.					

SECTION B (to be filled in by Medical Officer/Paediatrician)
39. Birth defects identified through instruments (Functional) (please tick √)
O a. Congenital deafness O b. Congenital vision defects O c. ROP O d. CHD
40. Birth defects identified through blood test (Metabolic) (please tick √)
A. IEM: O 1. CH O 2. CAH O 3. G6PD O 4. Others B. Haemoglobinopathy: O 1. Thalassemia O 2. Sickle Cell Disease 03. Others

41. Details of Congenital birth defects detected through instruments or blood test (functional and metabolic)

S.N	Name	Description of the anomaly	Age at diagnosis	Code- ICD10	Confirmed or suspected
a.	*Congenital deafness*				
b.	*Congenital vision defect*				
c.	*ROP*				
d.	*Congenital Heart Disease*				
e.	*IEM*				
f.	*Haemoglobinopathy*				

42. Photographs/reports attached: O Yes O No

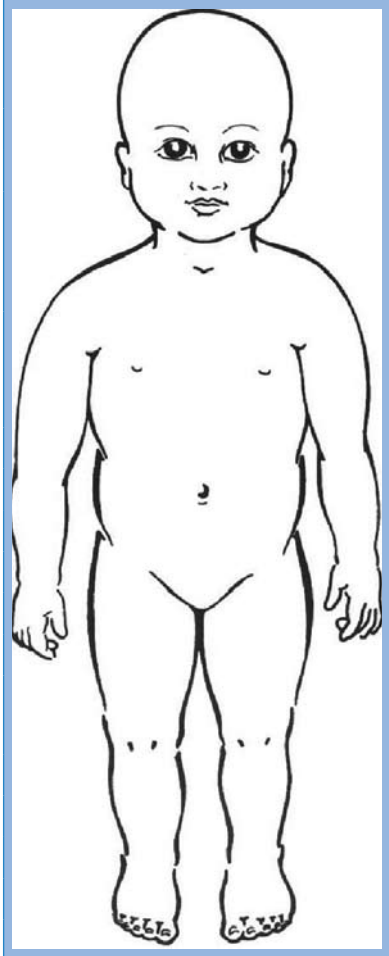
43. Investigation details

S.N	Relevant test	Result	Findings (if abnormal)
a.	Karotype/infantogram/ECG/US abdomen/Brain MRI/Any other	O N O AbN O Pending	
b.	Blood test, for O CH O CAH O G6PD O SCD O Others	O N O AbN O Pending	
c.	BERA, Fundoscopy etc.	O N O AbN O Pending	

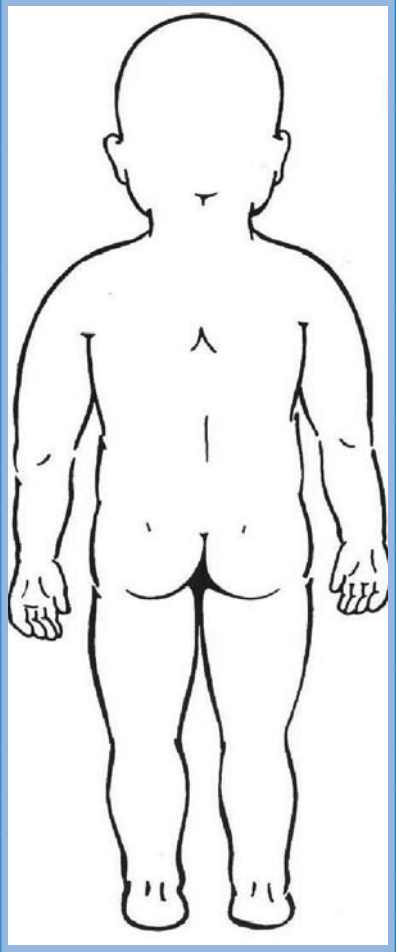
44. Diagnosis

Anomaly	Syndrome	Provisional diagnosis	Complete diagnosis
Notifying person	Designation and contact details:	Facility referred	Provisional diagnosis status O Confirmed O Suspected

BIRTH DEFECTS ENLISTED FROM HEAD TO TOE



Body Part	Visible
Head & Spine	• Neural Tube Defect- Anencephaly, Encephalocele, Spina Bifida • Arhinencephaly/ Holoprosencephaly • Hydrocephalus • Microcephaly
Face	Asymmetrical/dysmorphic
Eye	
• Eyelid	Hemangioma, Ptosis (partial closure of the lid), Coloboma (gap)
• Eyeball	Anophthalmos, Microphthalmos
• Inside eye	Corneal clouding, Coloboma of Iris, Congenital cataract, Congenital glaucoma
Ear	Anotia, Microtia, Low set ears
Mouth and Lips	Cleft lip, Cleft palate, both
Abdomen and Anus	
• Scaphoid & sunken	Diaphragmatic hernia
• Distended	Congenital intestinal obstruction
• Herniation of gut	Gastroschisis, Omphalocele
• Anus	Imperforate anus, anorectal malformation
Urinary tract	
• Bladder	Bladder exstrophy
• Urinary stream	Posterior urethral valve
• Lower Abdomen	Prune belly
Genitalia	
Ambiguous	Indeterminate sex
Urethral opening	Hypospadias
Limb	Limb reduction Defects, Club foot, Polydactyly, Syndactyly, Oligodactyly
Chromosomal	Down Syndrome Others
Others	



ANNEXURE - VIII
VARIABLES AND CODING INSTRUCTIONS OF CASE RECORDING FORM

Variable Number	Variable Name	Explanation and instructions	Digits	Code
1	**Location of reporting**	1 = State 2 =District 3 = Block	1	
2	**Source of Identification**	*Name of the Health Facility from where the fetus or new born with a congenital abnormality was identified* Number 01 = Sub-centre (SC) 02 = PHC 03 = Block Primary Health Centre (BPHC)/CHC 04 = Rural Hospital (RH) 05 = Sub-divisional Hospital (SDH) 06 = District Hospital (DH) 07 = Medical College (MC) 08 = Any other government institution 09 = Tertiary centre 10 = Any other	2	Code 01 to 13
3	**Reporting Month**	01 to 12 (January to December)	2	2 digits
4	**Reporting Year**	04 Digits, e.g. 2015	4	4 digits
5	**Date of Identification or Diagnosis** of Congenital Anomaly	Date on which the baby was first suspected or recognized as being malformed, even if the detailed diagnosis is not available. Please enter date as numeric string, 010215	6	Day Month Year 99 = Not known for day 99 = Not known for month 77 = Not known for year

Variable Number	Variable Name	Explanation and instructions	Digits	Code
6	**Time of Identification** for Congenital Anomaly	When the baby was first suspected to be malformed	1	1 = At birth 2 = < 1 month 3 = 1-12 months 4 = 1-6 years (beyond 12 months completed up to 6 years) 5 = Over 6 years 6 = Prenatal diagnosis 7 =At spontaneous Abortion or termination 8 = At autopsy 9 = Unknown
7	**Birth date**	Date of birth Please enter dates as a numeric string, not in date format (e.g. Do not use 01/02/89 or 01-02-89), instead use 010289 (DDMMYY)	6	Day, month, year 99 = unknown for day and month DO NOT TRANSMIT RECORDS IF YEAR OF BIRTH IS UNKNOWN
8	**Outcome of delivery**	O Live Birth O IUD/Still birth, <20 wks Type of birth The distinction between live births, stillbirth and spontaneous abortion should be followed. **Stillbirth:** Stillbirth is a baby borne without any signs of life after completing 20 weeks of pregnancy or 140 days or fetus weighing more than 500 gms. **MTP following anomaly:** When a pregnancy is terminated where the length of the pregnancy has not exceeded 20 weeks and there is a substantial risk if the child were born it would suffer from physical or mental abnormalities to be seriously handicapped. This is done as per the MTP Act 1971. **Spontaneous Abortions with anomaly:** If the fetus died spontaneously in utero, before completing 20 weeks of pregnancy (the length of the pregnancy has not exceeded 20 weeks) or weighing less than 500 gms.		1 = live birth 2 = stillbirth
9	**No. of babies**	Number of babies / fetuses delivered. Fill out a separate form for each malformed baby/ fetus in a multiple set. If the co-twin is not malformed, record its sex and whether it was live born or stillborn in the space for comments on the form of the malformed case.	1	1 = Singleton 2 = Twins 3 = Multiple (more than 2)

Variable Number	Variable Name	Explanation and instructions	Digits	Code
10	**Birth weight**	Birth Weight Give weight in grams If less than 1,000 grams, e.g. 540 gm code as 0540 If the fetus or neonate was stillborn, document the weight in grams.	4	I. Extremely low birth weight (ELBW) of less than 1000 gms or up to 0999 gms II. Very low birth weight (VLBW) – 1000 to 1499 gms III. Moderately Low Birth Weight (MLBW) from 1500 to 2499 gms
11	**SEX**	Indicate chromosomal sex, if known, in case of ambiguous genitalia with unknown or abnormal sex chromosome complement.	1	1 = Male 2 = Female 3 = Ambiguous 4 = Intersex
12	**GEST. AGE**	Length of gestation in completed weeks after first day of last normal menstrual period (LMP)	2	99 = not known
13	**LMP** (First day of the last normal menstrual period or on a first trimester sonogram)	**If LMP available and certain, calculate gestational age from date of LMP to date of delivery.** If LMP is available but uncertain, give the corrected gestational age by clinical ascertainment or other means. If LMP is not available, give the estimated gestational age by clinical ascertainment or other means	2	
14	**Birth Asphyxia**	1= yes 2 = no	1	
15	**Autopsy shows Birth Defect**	If IUD or Still birth autopsy done A = Yes. Autopsy done in a neonate / fetus born as IUD or Still birth B = Autopsy not done in IUD or Still birth	Alfa numerical	A0 = Autopsy done in Still birth but no Congenital Anomalies found A1 = Autopsy done in Still birth but with Congenital Anomalies
16	**Status of Induction / Augmentation**	0 = Not induced with Oxytocin or Misoprostol 1 = Induced or Augmented with Oxytocin 2 = Induced or Augmented with Misoprostol		
17	**Place of birth** **O Home** **O Institution**	State Dist Block Municipality	Alfa numerical	1 = home 2 = Institution 3 = State 4 = Block 5 = Municipality Villagel 99 = Not known
18 a	**MCTS NUMBER**	Mother and Child Tracking System is an IT enabled application which will facilitate monitoring of universal access to maternal and child health services. It is a name based tracking system launched by the Government of India to track all pregnant women and children.	16	Up to 16 digits.

Variable Number	Variable Name	Explanation and instructions	Digits	Code
18b	**UNIQUE ID** *Local ID* *States / District / Block / Year / Serial No.*	Each case has a unique identification. This is a maximum of 13 characters long. Consisting of numbers, letters, or both. ID numbers should not repeat themselves in different years.	13	Up to 13 digits 2 digits state code 2 digits district code 3 digits block PHC / CHC code 2 digits year code 4 digits serial code *From 1st April each year, the codes to be given a fresh starting from 0001.*
18c	**Mother's Aadhar No.**		12	
18 d	**Mobile No.**		10	
19	**Child's Name**	Alphabets		
20	**Mother's Name**	Alphabets		
21	**Mother's Age**	Years completed at the time of the birth of the child		
22	**Father's Age**	Years completed at the time of the birth of the child		
23	**Caste**	1 = Schedule Caste 2 = Schedule Tribe 3 = Other Backward Class 4 = General	1	1 SC 2 ST 3 OBC 4 Others
24	**Permanent Address** **State/District/ Block / H.No**	Address should be such the chance of tracking of the individual child is done easily. In case, the parents are migrant laborers, the place of delivery should be indicated. Mention the State, district and block or State and City.	Alpha	
25	**Folic Acid Intake** (Peri-conceptional Period)	Periconceptional Period: period from before conception to early pregnancy.	1	1 = Yes 2 = No 9 = Unknown
26	**H/o serious Maternal Illness**		1	1 = Yes 2 = No 9 = Unknown
27	**H/o Radiation Exposure**		1	1 = Yes 2 = No 9 = Unknown
28	**H/O Substance Abuse**		1	1 = Yes 2 = No 3 = Habitual exposure of smoking 4 = alcohol 5 = narcotic drug use 9 = Unknown

Variable Number	Variable Name	Explanation and instructions	Digits	Code
29	**Parental Consanguinity** *(Parents of fetus / neonate related)*	For example: (a) 1st cousin is the child of one's uncle and aunt. *(Uncle is the brother of your father or mother. Aunt is the sister of your father or mother.)* (b) 2nd cousin is the grand child of one's great uncle and great aunt or (c) If two people are first cousins, the children of each of them will be second cousins. *(Great uncle is the brother of your grandfather or grandmother & great aunt is the sister of your grandfather or grandmother.)* (d) Aunt – Nephew (e) Uncle – Niece (f) Other	1	1 = Yes 2 = No 9 = Unknown
30	**Assisted Conception**	Assisted Conception Assisted conception" means a pregnancy resulting from any intervening medical technology, whether in vivo or in vitro, which completely or partially replaces sexual intercourse as the means of conception IVF = In vitro fertilization GIFT – Gamete intra fallopian transfer	1	0 = No 1 = Induced ovulation only 2 = Artificial insemination 3 = IVF 4 = GIFT 8 = Other 9 = Not known
31	**Immunization History (Rubella)**		1	1 = Yes 2 = No 9 = Unknown
32	**Drugs during first trimester**: i.e. from the first day of LMP to 12th week of gestation	00 – 19 88, 98, 99 00 = No drugs 01 = Atropinics and antispasmodics 02 = Anaesthetics, local and general 03 = Hypnotics, sedatives and psychotropics 04 = Antiepileptics 05 = Analgesics, antipyretics and anti-inflammatory agents 06 = Histamine antagonists 07 = Antiashthmatic agents including methylxanthines 08 = Antiarhythmic and antihypertensive agents 09 = Diuretics 10 = Tocolytics 11 = Antiseptics, antibiotics, antiviral, antiparasitic and antifungal agents 12 = Antiproliferative & immunosuppressive agents 13 = Anticoagulant antithrombotic and thrombolytic drugs	2	

Variable Number	Variable Name	Explanation and instructions	Digits	Code
32	**Drugs during first trimester**: i.e. from the first day of LMP to 12th week of gestation	14 = Thyroid and antithyroid drugs 15 = Oestrogens, progestins and androgens, including oral contraceptives 16 = Adrenocortical steroids 17 = Insulin and oral hypoglycemic agents 18 = Vaccines 19 = Vitamins and minerals 88 = Other 98 = Drug(s) taken but no information available 99 = Not known	2	
33	**Anomaly in previous pregnancies**	1 = Yes 2 = No 9 = Unknown		
34	**Total number of previous abortions**	No. of previous abortions (exact numbers)	2	01 = One 02 = Two 03 = Three 04 = Four 05 = Five 06 = 6+ 99 = Unknown
35	**Total No. of previous Still birth**	No. of previous still birth (exact numbers)	2	01 = One 02 = Two 03 = Three 04 = Four 05 = Five 06 = 6+ 99 = Unknown
36	**Head Circumference**	Head Circumference in centimeters	2	99 = Not measured
37	**Type of Defects**	1 = Single 2 = Multiple	1	
38	**Visible Birth Defects (Head and spine; Face, eyes, ears, mouth and lips; Abdomen and anus; Genitalia; Urinary tract; Limbs (upper and lower)**	Type & Site, Description of the anomaly, Age at diagnosis, Code- ICD10, Confirmed or suspected		

ANNEXURE - IX
CASE DEFINITIONS AND GLOSSARY OF TERMS

Case Definition	
Birth Defect or congenital anomaly or congenital malformation	Abnormal health conditions which are present since birth but not necessarily manifest at Birth are referred to as Birth Defects. 'Congenital abnormality', 'congenital anomaly', and 'congenital malformation' are terms often used as synonyms for 'Birth Defect'. However, the word 'congenital' may describe any condition present at birth, regardless of its etiology or timing of occurrence. *The term Birth Defect encompasses a diversity of conditions including physical malformations such as cleft lip or Palate, chromosomal abnormalities such as Down syndrome, functional defects including sensory deficits such as congenital deafness and congenital cataract, metabolic defects including inborn error of metabolism or Haemoglobinopathies, neurodevelopmental disorders, and complications related to prematurity such as retinopathy of prematurity and low birth weight, among others. (National Birth Defects Prevention Network, CDC).*
Comprehensive Newborn screening	Includes screening for Birth Defects that includes *physical malformations, chromosomal abnormalities, functional defects, metabolic defects, neurodevelopmental disorders and complications related to prematurity. Screening is done at 4 levels, Delivery points, Birth to 6 weeks through ASHA, 6 weeks to 6 years at Anganwadi centers and 6 years to 18 years at schools. Both active and passive screening is included.*
Definition of General Terms	
Major anomaly	A congenital abnormality that requires medical or surgical treatment, has a serious adverse effect on health and development. Individual major anomalies occur in less than 1 percent of the population. Together, they are seen in approximately 3 percent of births. Examples include cleft lip and tracheo-esophageal fistula.
Minor anomaly	A congenital abnormality that does not require medical or surgical treatment, does not seriously affect health and development, and does not have significant cosmetic impact. Individual minor anomalies generally occur in less than 4 percent of the population. *The presence of multiple minor anomalies in the same child* ***may indicate the presence of an undiagnosed major anomaly, syndrome, or functional deficit.***
Normal variant	A minor anomaly that occurs in approximately 4 percent or more of the population. Examples of normal variants include webbing of the second and third toes and a single umbilical artery in an otherwise normal infant.
Definition of terms Related to the Formation of Major Anomalies	
Malformation	A major anomaly that arises **during the initial formation of a structure**, i.e. during organogenesis. For most organs, **this occurs during the first eight weeks** after fertilization. The resulting structure may be abnormally formed, incompletely formed, or may fail to form altogether. Examples of malformations include spina bifida and hypoplastic left heart. The term 'congenital malformation' is also used more broadly to indicate any major anomaly.
Disruption	A major anomaly that results from **alteration of a structure after its initial formation**. The resulting structure may have an altered shape and configuration, abnormal division or fusion of its component parts, or loss of parts that were previously present. Examples of disruption defects include intestinal atresia and possibly gastroschisis.
Deformation	A major anomaly that results from molding of part of a structure, usually over a prolonged time, by mechanical forces after its initial formation. Examples of forces that may lead to a deformation include oligohydramnios (diminished amniotic fluid) and intrauterine crowding in twin, triplet, or higher order pregnancies. Examples of deformations include the compression (Potter's) facies seen with bilateral renal agenesis and some instances of clubfoot.
Definition of terms Related to Patterns of Multiple Anomalies Occurring in a Single Child	
Syndrome	A pattern of anomalies that form a specific diagnosis for which the natural history and recurrence risk are usually known. Use of the term 'syndrome' implies that the anomalies have a common specific etiology. Examples include Beckwith-Weidemann syndrome and Rubinstein-Taybi syndrome.

Sequence	*A pattern of anomalies that results from a single primary anomaly or Mechanical factor*. **The presence of the initial anomaly or factor leads to one or more secondary anomalies**, which may then lead to one or more tertiary anomalies in cascade fashion. Examples include Robin sequence (micrognathia, posterior displacement of the tongue, cleft soft palate) and the oligohydramnios, or Potter, sequence (pulmonary hypoplasia, flattened facies, abnormal positioning of the limbs).
Association	*A non-random pattern of anomalies that occur together more frequently than expected by chance alone*, but for which no etiology has been demonstrated. Examples include VACTERL association (Vertebral, Anal, Cardiac, Tracheo-Esophageal, Renal, and Limb anomalies) and CHARGE association (Colobomas, Heart defects, choanal Atresia, Retarded growth and development and/or central nervous system anomalies, Genital anomalies and/or hypogonadism, Ear anomalies and/or deafness). Use of the term 'association' does not indicate that a specific diagnosis has been made.
Terms Related to Tissue and Organ Formation	
Agenesis	Failure of an **organ** to form.
Dysgenesis	Anomalous or disorganized formation of an **organ.**
Aplasia	Absence of a **tissue or organ** due to lack of cell proliferation.
Dysplasia	Disorganized cell structure or arrangement within a **tissue or organ**.
Hypoplasia	Undergrowth of a **tissue or organ** due to insufficient proliferation of normal cells.
Hyperplasia	Overgrowth of a **tissue or organ** due to excess proliferation of otherwise normal cells.
Terms Related to the Timing of Gestation and Delivery	
Embryonic period	**The first eight weeks** after fertilization, during which most, but not all, organs are formed.
Fetal period	The period from the ninth week after fertilization through delivery.
Neonatal (Newborn) period	The first 28 days following delivery of a live-born infant.
Prenatal period	Before delivery.
Perinatal period	Before, during, or after delivery. The exact time period may vary **from 20 to 28 completed weeks of gestation through 7 to 28 days after delivery**, depending on the context in which the term is used.
Terminology Related to Pregnancy Outcome	
Live birth	**Spontaneous delivery of an infant that exhibits signs of life**, including a heartbeat, spontaneous breathing, or movement of voluntary muscles. Transient cardiac contractions and fleeting respiratory efforts or gasps are not necessarily considered signs of life by all programs.
Fetal death (stillbirth)	**Spontaneous delivery of an infant or fetus at 20 weeks or greater gestation that does not exhibit signs of life**. Transient cardiac contractions and fleeting respiratory efforts or gasps are not necessarily considered signs of life by all programs. A late fetal death is a fetal death that occurs at 28 weeks or greater gestation.
Spontaneousabortion (miscarriage)	Spontaneous delivery of a fetus at less than 20 weeks gestation.
Induced abortion (elective termination)	The purposeful interruption of pregnancy with the intention other than to produce a live birth and which does not result in a live birth.
Term infant	An infant born after 37 completed weeks and before 42 completed weeks of gestation.
Preterm infant	An infant born before 37 completed weeks of gestation.
Post term infant	An infant born after 42 completed weeks of gestation.
Low birth weight	Birth weight less than 2,500 grams, regardless of gestational age
Very low birth weight	Birth weight less than 1,500 grams, regardless of gestational age.
Extremely low birth weight	Birth weight less than 1,000 grams, regardless of gestational age.
Neonatal death	Death of a live-born infant within the first 28 days after birth.
Early neonatal death	Refers to death during the first 7 days.
Late neonatal death	Refers to death after 7 days but before 29 days.
Infant death	Death of a live-born infant before 12 months of age.

ANNEXURE - X
REFERRAL FORM

Referral Form
Include any congenital abnormality identified / strongly suspected

Date & Time of Referral: Day ☐☐ Month ☐☐ Year ☐☐☐☐ Time: Hour ☐☐ Min ☐☐ AM/PM ☐☐	**Age of the child on the day of referral:** Below 1 Month ☐ 1 Month to 6 Months ☐ 6 Months to 1 Year ☐ Beyond 1 Year ☐
Name of the Medical Facility referred to :	
Contact Person or department referred to :	

Status of Referral Visit : Tick as appropriate	☐ Urgent:* less than 24 hrs.√	☐ Semi urgent : within 2 weeks √	☐ Routine : within 30 days √

DESCRIPTION

Type of birth defect : Tick as appropriate	☐ Single (isolated) birth defect	☐ Multiple birth defects	Syndrome suspected : ☐ No ☐ Yes

Description of Birth Defect (S): Please describe each birth defect in as much details as possible. If syndrome suspected add details. Add confirmation document when available	ICD-10 codes
Head and spine :	
Face, Eye , Nose, Ears , Mouth including Lip and Palate :	
Chin, Neck and Clavicle :	
Chest , Lung and Heart :	
Abdomen including anus :	
Genitalia :	
Kidney, Ureters and Urinary Bladder	
Limbs:	
Hip:	
Chromosomal disorder :	
Neurodevelopmental disorder :	

Immediate Medical Issues that need to be addressed urgently :

1.
2.
3.

Affected newborn/ infant / child : please fill in one form per affected child

Mother's name :		**Child's name :**		☐ **Child** √ **or** ☐ **Mothers** √ ADHAR or MCTS / hospital ID number ☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐
State :	**District :**	**Block :** **Municipality:**	**Place of birth:** ☐ Home ; ☐ Institution ***Facility code no.**	**Phone no :** ☐☐☐☐☐☐☐☐☐☐
Birth asphyxia ☐ Yes ☐ No	**Date of Birth (NB)** D D M M Y Y Y Y	**Sex :** ☐ Male ☐ Female ☐ Ambiguous		**No. of Babies:** ☐ Single ☐ Twins ☐ Multiple If multiple : Order : 1st / 2nd /3rd /4th

Referring provider details

Name: ……	Designation: ……
Name of the medical facility: ……	Contact Phone ……E-mail ……

√ Tick below for the specialty/specialist to which referral is being made

Pediatric Medicine	Neurosurgery	Pediatric Surgery	Genetics or pediatrician trained in medical genetics	Ophthalmologist trained in Pediatrics.	Optometrist trained in Pediatrics.	Ophthalmologist trained in Retinal (ROP)	ENT specialist Trained in Pediatrics	Audiologist trained in Pediatrics	Plastic Surgery
Pediatric Dentist	Pediatric Cardiologist or Pediatrician trained in Cardiology	Pediatric Cardiothoracic Surgeon	Radiologist	X-Ray Technician	Sonologist	Pediatric Endocrinologist	Urosurgeon Trained in Paed. or Pediatric surgeon	Gynecologist Trained to manage an infant	Orthopedic surgeon
Lactation Management unit	Pediatric Neurologist	Occupational Therapist	Physiotherapist Trained in pediatrics	Plaster technician at Club foot clinic / DEIC	Cleft Lip Cleft palate Craniofacial center / DEIC	Downs syndrome at DEIC	Dermatologist Trained in Pediatrics	Child Psychiatrist	Nutrition Support Center (NRC)

*** Facility code no: 01) Sub Centre, 02) PHC, 03) CHC/BPHC, 04) Rural Hospital, 05) Sub divisional Hospital, 06) District Hospital, 07) Medical College Hospital, 08) Other Govt. Hospital 09) Tertiary center 10) any other**

Head ☐√ and Spine ☐√

Head

- Neural Tube defect (Q:00,01,05): ☐√
- Anencephaly(NTD) (Q00): ☐√
- Encephalocele (NTD)(Q01): ☐√
- Myelomeningocele (NTD)(Q05): ☐√
- Microcephaly (Q02): ☐√
- Congenital Hydrocephalus(Q03):☐√
- Arhinencephaly (Q04.1): ☐√
- *Holoprosencephaly (Q04.2): ☐√
- Macrocephaly (Q75.3): ☐√
- *Craniosynostosis (Closed fontanelle and fused suture): (Q75.0): ☐√

Spine

- Abnormal curvature of spine ; Congenital scoliosis: (Q67.5): ☐√
- Spina bifida occulta (Tuft of hair or dimple along intact spine) : (Q76.0): ☐√

may require Imaging

Face ☐√,Ear ☐√, Mouth ☐√,Chin ☐√,Neck ☐√, Clavicle ☐√

Face

- Facial asymmetry; Asymmetry on crying (Q67.0): ☐√
- Dysmorphic features; Down syndrome(Q90):☐√

Ear

- Absence of external ear; anotia (Q16): ☐√
- Microtia (Q17.2): ☐√
- Misplaced ear (Low-set ears)(Q17.4): ☐√
- Congenital Deafness (H90): ☐√

Mouth

- Cleft Lip (Q36): ☐√
- Cleft Palate (Q35): ☐√
- Cleft lip with Cleft Palate (Q37): ☐√

Chin, Neck, Clavicle

- ***Chin***: Small chin or microganthia (M26.0): ☐√
- ***Chin***: Receding chin or retoganthia (M26.19): ☐√
- ***Chin***: Pierre-Robin sequence; Triad; Receding Chin + backward displacement of Tongue+ posterior cleft palate(Q87.0): ☐√
- ***Neck***: Webbing of neck (Q18.3): ☐√
- ***Neck swelling***/mass :
  - ***a.*** cystic swelling on the lateral side of the neck: Cystic Hygroma (D 18.1) ☐√; Branchial cleft cysts (Q 18.0) ☐√;
  - b. cystic swelling on the midline of neck : Thyroglossalduct cysts (Q 89.2) ☐√;
  - ***c.*** solid swelling in the neck: midline: thyroid swelling (E 03) ☐√, lateral side :congenital torticollis(Q 68.0) ☐√
- ***Clavicle***: Absence of Clavicles, malposition of scapula (Q74.0) ☐√

Eyes ☐√

- ***Eyelid***: Congenital ptosis(Q10) ☐√
- ***Eyelid***: Coloboma of eyelid(Q10.3) ☐√
- ***Eyelid***: Hemangioma (eyelid)(D18.01) ☐√
- ***Facial Mark***: Port wine stain(Q82.5) ☐√

Eyeball size: absence or abnormally small or abnormally large Eye ball

- Anophthalmos: (Q11.1) ☐√
- Microphthalmos: (Q11.2) ☐√
- Congenital glaucoma(Q15.0): ☐√

Folds on the inner or ***medial canthus***

- Epicanthic fold (Q10.3): ☐√

Inside the eye: cornea, pupil, iris, lens, retina

- ***Corneal*** opacity (hazy, dull)(Q13.3): ☐√
- ***Pupils*** unequal size & shape(Q13.2): ☐√
- ***Iris***: Coloboma of iris(Q13.0): ☐√
- ***lens***: Congenital Cataract(Q12.0): ☐√
- ***Retina***: Retinopathy of prematurity(H35.1): ☐√
- ***White pupillary reflex seen*** on torch examination ☐√
  - ***a.*** seen in corneal opacity, e.g., cataract ☐√
  - ***b.*** seen in Retinal pathology, e.g.,
  - *Retino Blastoma (RB) ☐√;
  - *Retinopathy of prematurity (ROP) ☐√;
  - *Persistent Hyperplastic Primary Vitreous, also referred as persistent fetal vasculature as it fails to regress(PHPV) ☐√

Chest wall ☐√; Respiratory ☐√ ; Cardiac ☐√

Chest wall & Respiratory

- Abnormality in chest wall:- size, shape, symmetry, number & position of nipples (Q67.6/67.7): ☐√
- Respiratory distress of newborn(P22): ☐√
- Apneic disorders(P28.4) : ☐√

Cardiac : Abnormal pulses:

- ***Point of maximum impulse or Apical Impulse :*** displaced on the right side
- ***Weak or absent pulse in all extremities*** (decreased cardiac output) + >60/breaths per minute can be due to cardiogenic shock (Q23.4): ☐√ Hypo plastic left heart syndrome- a condition caused by underdevelopment of the whole left half of the heart.)
- ***Weak or absent only femoral pulse*** : Coarctation of Aorta(Q25.1) : ☐√ (Birth defect with congenital narrowing of Aorta)
- ***Blue lips*** not improving with oxygen therapy associated with nonsignificant respiratory distress (Q24.9): ☐√
- ***Positive pulse Oximetry***: ☐√ Spo2 <95%

Genitalia ☐√; Male genitalia ☐√;Female genitalia ☐√; Abdomen ☐√ and Anus ☐√

Genitalia

- Micropenis: less than 2.5 cm, stretched length(Q55.62): ☐√
- Bilateral undescended Testes(Q53.2): ☐√
- Testicular torsion(N44): ☐√
- Unequal scrotal size or scrotal discoloration: ☐√
- Congenital Hydrocele(fluid filled sac or scrotum)(P83.5): ☐√
- Absence of vaginal opening(Q52.0): ☐√
- Ambiguous genitalia (Q56.4): ☐√

Abdomen : Abdomen shape either distended or Scaphoid

- Distended i.e. Congenital intestinal obstruction (Q41-42): ☐√
- Abnormal abdominal mass : ☐√
- Scaphoid along with respiratory distress(Congenital Diaphragmatic Hernia or CDH) (Q79.0): ☐√

Defect in Abdominal wall

- Abdominal wall: Omphalocele/Exomphalos(Q79.2) : ☐√
- Abdominal wall: Gastroschisis (Q79.3) : ☐√
- Inguinal hernia/swelling(K40): ☐√
- Umbilical hernia/swelling(K42.9): ☐√

Anus

- Imperforate anus with or without fistula**(Q42.0-42.3): ☐√
- Abnormal position of anus(Q43.5): ☐√

****Fistula is suspected if meconium seen in urine or through vagina in females***

Urinary tract ☐√

- Bladder Exstrophy (Q64.0-Q64.1): ☐√
- Posterior Urethral Valve(PUV)(Q64.20): ☐√ PUV: Urine flow drop by drop instead of continuous flow & bladder distended even after newborn has passed urine.
- *PUJO: Pelvic Ureteric Junction obstruction is a partial or intermittent total blockage of the flow of urine that occurs where the ureter enters the kidney. PUJO obstruction is the most common pathologic cause of antenatal detected Hydro nephrosis (Q62.11) : ☐√
- Congenital Hydronephrosis(Q62.0) : ☐√
- Hypospadias (hypo=under)(Q54) : ☐√
- Epispadias(Epi=above)(Q64) : ☐√
- Prune Belly syndrome (Q79.4) : ☐√
- Potters syndrome (Q60.6) : ☐√

****For the first 24 hours after birth, the infant's serum chemistries are the same as the mother i.e. newborn Urea and creatinine to be done after 24 hours***

Upper limb ☐√

- Complete absence of the whole of the upper limb i.e. arm/forearm, hand including digits (Q71.0): ☐√
- Absence of arm & forearm with hand present (Q71.1) ☐√
- Absence of forearm & hand(Q71.2): ☐√
- Absence of hands and fingers (Q71.3): ☐√
- Longitudinal reduction defect of radius i.e. Absence or hypoplasia of radius bone (Q71.4) : ☐√
- Longitudinal reduction defect of Ulna i.e. Absence or hypoplasia of Ulna bone Q71.5) : ☐√
- Lobster claw hand (Q71.6) ☐√
- * Single transverse crease: ☐√

Lower limb ☐√

- Complete absence of the whole of the lower limb i.e. thigh/leg/ foot(Q72.0): ☐√
- Absence of thigh & lower leg but foot present(Q72.1): ☐√
- Absence of both lower leg & foot (Q72.2): ☐√
- Absence of foot and toe fingers (Q72.3): ☐√
- Congenital shortening of lower limbs (Q72.81) : ☐√
- Lobster split foot (Q72.7): ☐√
- Fusion or Webbing between adjacent fingers or toes or Syndactyly (Upper limb/lower limb)(Q70): ☐√
- Extra digits or Polydactyly of fingers/thumb/toes (Q69) : ☐√

Chromosomal disorder ☐√ NDD ☐√

Look for any dysmorphic feature. Look at the face for upward slanting eyes, epicanthic fold, flat nose, small ears, small mouth, single

Palmar crease and increase gap between the first and second toe.

- Presence of chromosomal disorder: ☐√
- Downs syndrome(Q90): ☐√

NDD (Neurodevelopmental disorder) ☐√

- Altered state of consciousness: ☐√

Cry

- Weak or absent cry : ☐√
- Inconsolable cry : ☐√
- Tonal abnormality : Increased tone or decreased tone (Floppy) ☐√
- Seizures: ☐√

ANNEXURE - XI

KNOW YOUR REFERRAL SERVICES : KYRS

Mapping of available services for facilitating appropriate referrals			
Health Conditions(Defects at Birth) managed under RBSK, free of cost :			
1) Neural Tube defect **2) Cleft lip & Palate** **3) Down syndrome** **4) Congenital cataract** **5) Congenital deafness**			**6) Congenital Heart disease** **7) Talipes (Club foot)** **8) Developmental Dysplasia of Hip** **9) Retinopathy of Prematurity**
NB: all other health conditions are managed free or cost under JSSK till 2 years of age			
Defects at Birth **Please Note (☐) indicates urgent referral, where attention is required in 24 hours** ☐ *Urgent:* less than 24 hrs.√* ☐ ☐ *Semiurgent : within 2 weeks √* ☐ ☐ *Routine : within 30 days √* ☐ **status of referral visit*			**Name and address of the Institution where services are expected to be available including contact person must be prepared, filled and made available at all delivery points (NB: Please indicate the specialty or super specialty, apart from the name of the institute and contact person).** Resouce mapping of the required services will enable to fill these forms
Head: Shape and symmetry	*☐☐☐	Specialty required for referral	**Name and address of the specialist including the institution**
○ ***Scalp swelling***	☐	○ Pediatrician	
○ **Herniation of the brain through a defect in the skull : Encephalocele**	☐	○ Neurosurgery or Pediatric surgery	
○ ***Absence of cranial vault :*** **Anencephaly** *Referring for surgery is of not much use as most children would die	☐	○ Pediatrician, especially for Counselling the family on folic acid	
Anterior and posterior fontanelle			
○ ***Closed fontanelles and fused sutures (craniosynostosis)***	☐	○ Neurosurgery or Pediatric surgery	
○ ***Enlarged, bulging or sunken fontanelle***	☐	○ Pediatrician	
○ ***Hydrocephalus***	☐	○ Neurosurgery or Pediatric surgery :	
Other Congenital Neurological Health condition			
○ ***Other congenital brain anomalies***	☐	○ Pediatrician	
Head circumference			
○ ***Microcephaly*** ○ ***Macrocephaly***	☐ ☐	**Pediatrician**	
Spine and Spinal column			
Abnormal swelling of the spine ○ ***Meningocoele*** ○ ***Meningomyelocoele***	 ☐ ☐	○ Neurosurgery or Pediatric surgery	
○ ***Abnormal curvature of spine*** ○ ***Tufts of hair or dimple along intact spine***	☐	○ Pediatrician	

Face			
○ ***Dysmorphic appearance of face***	☐	○ Pediatrician/Geneticist	
○ ***Asymmetry on crying***	☐		
Eye			
○ ***Eyelid : Swelling, drooping or gap in the eye lid***	☐	○ Pediatric Ophthalmologist ○ Pediatric Optometrist ○ **Opthalmic surgeon** trained for ROP ○ Pediatrician trained to treat purulent conjunctivitis	
○ ***Facial marks near the eye : Port wine stain***	☐		
○ ***Eyeball : Abnormally small eye or absent eye***	☐		
○ ***Eye Position in relation to the nasal bridge :Upward slant/downward slant/epicanthic fold***	☐		
○ ***Cornea:Hazy, dull cornea, opacity***	☐		
○ ***Pupil : Pupils unequal, dilated or constricted or gap in the pupil(coloboma)***	☐		
○ ***Lens: Congenital cataract***	☐		
○ ***Cornea opacity with tearing: Congenital glaucoma***	☐		
○ ***Conjuctiva : Purulent conjunctivitis***	☐		
○ ***White reflex through a torch***	☐		
○ ***Retinopathy of prematurity***	☐		
Ears			
○ ***Abnormal shape of ear or abnormal palcement of ear or absent external ear***	☐	○ Pediatric ENT specialist	
○ ***Family history of deafness or suspected deafness***	☐	○ Pediatric Audiologist	
Mouth and Lips			
○ **Cleft lip/plate : craniofacial abnormality** Cleft lip is repaired between 6-12 weeks after birth. Cleft palate is repaired between 12-18 months but initially a dental surgeon can occlude the opening by preparing an Obturator	☐ ☐	○ Plastic surgery or Pediatric surgery or Cleft lip/palate center ○ Dental surgeon can help in *application of Obturator in cleft palate*	
○ **Blue discoloration of lips : Congenital heart disease**	☐	○ Pediatrician ○ Pediatric cardiology ○ Pediatric cardiac surgery	
Nose			
Non-patent nares (bilateral nasal obstruction)	☐	○ Pediatric ENT specialist	
Chin , Neck and clavicles (collar bone)			
○ **Receding chin or lower Jaw : Retrognathia** ○ **Small chin or lower jaw : Micrognathia**	☐ ☐	○ Pediatrician ○ Pediatric surgeon ○ Radiologist	
○ **Neck webbing** ○ **Neck Masses : not obstructing the airway**	☐		
○ **Swelling in neck with obstruction of airway**	☐		
○ **Absence of clavicles**	☐		

Chest wall , Respiratory and Cardiac			
○ **Abnormality in chest size, shape and symmetry**	☐	○ Pediatrician	
○ **Abnormality in number and position of nipples**	☐		
○ **Respiratory distress of newborn : Urgent X-ray chest**	☐	○ Pediatrician	
○ **Apneic episodes : Very critical**	☐		
○ **Weak or absent femoral pulses**	☐	○ Pediatrician ○ Pediatric cardiology ○ Echocardiography (sonologist trained in newborn ECHO)	
○ **Weak or absent pulse in all extremities**	☐		
○ **Blue lips**	☐		
○ **Positive pulse Oximetry (if done)** Spo2 <95%	☐		
Abdomen and anus			
Abnormality in Shape and symmetry		○ Pediatric surgeon trained in: a. Neonatal gastro surgery b. Neonatal Anorectal surgery c. Neonatal Thoracic surgery ○ Pediatric anesthetist	
○ **Abdominal swelling: congenital intestinal obstruction: X-ray abdomen**	☐		
○ **Abdominal scaphoid with respiratory** ○ **Distress: X-ray abdomen**	☐		
○ **Bilious vomiting: X-ray abdomen**	☐		
○ **Abnormal mass in the abdomen**	☐		
Defect in the abdominal wall			
○ **Exomphalos** ○ **Gastroschisis**	☐ ☐		
Umbilicus including number of arteries			
○ **Less than 3 umbilical vessels**	☐		
○ ***Anus Position : abnormal***	☐		
○ ***Anal Patency : absent***	☐		
○ **No meconium passed within 24 hours**	☐	○ Pediatrician for treating jaundice	
○ **Abnormal position of anus**	☐		
○ **Absence , imperforate anus**	☐		
○ **Jaundice < 24 hours of age or on palms and soles**	☐		
Male and female genitalia			
Male genitalia : Penis including foreskin; Testes (confirm present bilaterally and position of testes) including any discoloration; Scrotal size and color; Other such as Hydrocele		○ Pediatrician trained in pediatric endocrinology ○ Pediatric surgeon required for treating *Testicular torsion*	
○ **Micropenis (stretched length less than 2.5cm)**	☐		
○ **Bilateral undescended testes**	☐		
○ **Testicular torsion**	☐		
○ **Unequal scrotal size or scrotal discoloration**	☐		

Female genitalia: Clitoris, Labia, Hymen & Urethral opening			
○ **Absence of vaginal opening**	☐	○ Gynecologist *(Specifically sensitized in handling newborns*	
○ **Pseudo menses (no referral required)**			
○ **Inguinal hernia/swelling (either male or female)** ○ **Umbilical hernia**	☐ ☐	○ Pediatric surgeon	
○ **Ambiguous genitalia (either male or female)**	☐	○ Pediatrician ○ Pediatric Endocrinologist	
Urinary tract			
Bladder wall: *check for intact or distended bladder. Check whether the newborn has passed urine and identify from where the urine comes out i.e. Urethral opening. Also Check for urinary stream in a male child* ***Bladder wall not intact- bladder Exstrophy**	☐	○ Pediatric Uro-surgeon ○ Pediatric surgeon Ultrasonologist ○ Radiologist	
○ **Posterior Urethral Valve (PUV)**	☐		
○ **Pelviureteric junction obstruction (PUJO)**	☐		
○ **Hypospadias**	☐		
○ **Epispadias**	☐		
○ ***Congenital Hydro nephrosis***	☐		
Limbs : Upper and Lower limbs			
Upper Limbs: Arm, Forearm, Hand, digits and palm; Lower Limbs: Thigh, Leg, Foot and toes		○ Pediatric orthopedic surgeon ○ Occupational therapist/ ○ Pediatric physiotherapist Plaster Technician ○ Club foot center	
○ **Absence of the whole or a part of the upperlimb (arm/forearm, hand)**	☐		
○ **Extra digits**	☐		
○ **Webbing of fingers**	☐		
○ **Single transverse crease**	☐		
○ **Absence of the whole or a part of the lowerlimb (*Thigh, Leg, Foot and toe*)**	☐		
○ **Extra digits in the foot**	☐		
○ **Missing digits in the foot**	☐		
○ **Clubfoot**	☐		
Hip			
Check symmetry of the legs , Skin folds over the buttocks , failure to abduct the hip & Risk factors for hip dysplasia: breech; females ; positive family history Congenital Hip dysplasia * USG most reliable below 4 months after 4 months X-ray Hip joint		○ Pediatric orthopedic surgeon ○ Occupational therapist ○ Ultrasonologist or ○ Radiologist	

Chromosomal			
Look for any **dysmorphic feature**. **Face** for upward slanting eyes, epicanthic fold, flat nose, small ears, small mouth, **Palm** for Single palmar crease and **Foot** for increase gap between the first and second toe. * Presence of chromosomal disorder * Downs syndrome	☐	○ Pediatrician ○ Down syndrome center ○ Pediatric physiotherapist/ Occupational therapist ○ Pediatric Geneticist	
Neurodevelopmental disorders			
○ Weak/irritable/absent cry	☐	○ Pediatrician ○ Pediatric neurologist ○ Occupational therapist/ Pediatric physiotherapist	
○ Absent reflexes	☐		
○ No response to crying even after consoling	☐		
○ Crying even when not touched or handled	☐		
○ Seizures	☐		
○ Altered state of consciousness	☐		

EXAMINATION OF THE NEWBORN FROM HEAD TO TOE FOR COMMON BIRTH DEFECTS

RBSK
RASHTRIYA BAL SWASTHYA KARYAKRAM
FROM SURVIVAL TO HEALTHY SURVIVAL

NATIONAL HEALTH MISSION
राष्ट्रीय स्वास्थ्य मिशन

GENERAL OBSERVATION : If present, refer

• Looks ill • Lethargic • Abnormal cry • Not feeding • **Colour of skin:** a) Pale b) Blue c) Yellow

Wash your hands, before touching the baby

1 HEAD AND SPINE

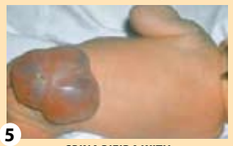
1. Size too large > 38 cms (full term)
2. Size too small < 32 cms (full term)
3. Absence of skull cap
4. Swelling or protruding of the brain
5. Abnormal swelling of the spine


1 *HYDROCEPHALOUS* Q03 *


2 *MICROCEPHALY* Q02


3 *ANENCEPHALY* Q00


4 *ENCEPHALOCELE* Q01


5 *SPINA BIFIDA WITH MENINGOMYELOCOELE* Q05

2 EYES, EARS, MOUTH AND LIPS

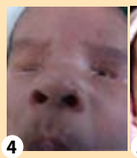
EYES

1. Eyelid – swelling
2. Eyelid – droopy
3. Gap in eyelid
4. Eyeball – absent
5. Eyeball – small
6. Inside the eye – corneal clouding
7. Inside the eye – opacity of lens/white reflex


1 *HAEMANGIOMA* D 18.01

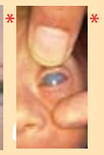

2 *PTOSIS* Q 10.0


3 *COLOBOMA OF EYELID* Q10.3


4 *ANOPHTHALMOS* Q11.1 *ONE EYE OR BOTH EYES*

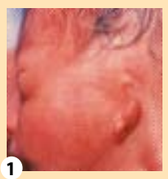

5 *MICROTHALMOS* Q11.2

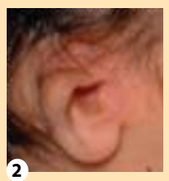

6 *CONGENITAL CATARACT* Q12.0 *

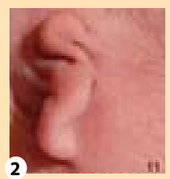

CONGENITAL GLAUCOMA Q15.0 *

EAR

1. Absent
2. Abnormal shape


1 *ANOTIA* Q 16.0


2 *MICROTIA II* Q 17.2


2 *MICROTIA III* Q 17.2

MOUTH

1. Cleft (split) lip
2. Cleft (split) palate
3. Cleft (split) lip and palate


1 *CLEFT LIP* Q 36

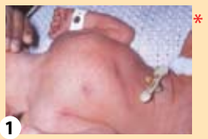

2 *CLEFT PALATE* Q 35

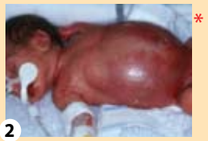

3 *CLEFT LIP & PALATE* Q37

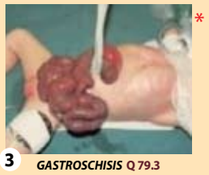
3 ABDOMEN AND ANUS

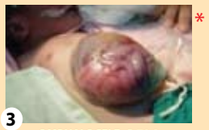
ABDOMEN

1. Scaphoid (sunken and concave) with respiratory distress: X-ray chest
2. Distended: X-ray abdomen
3. Wall defect- gap with herniation of the gut


1 *DIAPHRAGMATIC HERNIA* Q 79.0 *

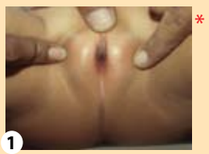

2 *INTESTINAL OBSTRUCTION* Q 41-42 *


3 *GASTROSCHISIS* Q 79.3 *


3 *OMPHALOCELE* Q 79.2 *

ANUS

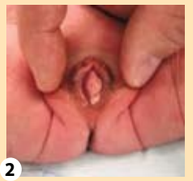
1. Absent/imperforate/ abnormally positioned

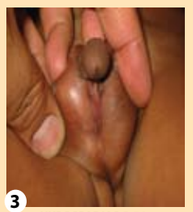

1 *IMPERFORATE ANUS/ANORECTAL ATRESIA AND STENOSIS WITH OR WITHOUT FISTULA* Q42.0-Q42.3 *

4 GENITALIA

1. Ambiguous genitalia
2. Vaginal opening absent
3. Urethral opening away from the tip of the penis – look where the urine comes out


1 *AMBIGUOUS GENITALIA* Q 56.4 *

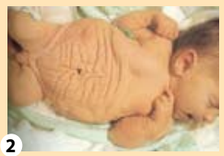

2 *VAGINAL AGENESIS* Q 52.0


3 *HYPOSPADIAS* Q 54

5 URINARY TRACT

1. Bladder – not covered
2. Wrinkled abdominal wall
3. Urinary stream – check if male child


1 *BLADDER EXSTROPHY* Q 64.0-64.1 *

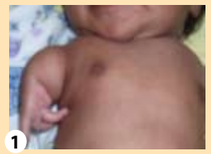

2 *PRUNE BELLY* Q 79.4

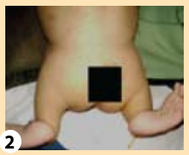

3 *POSTERIOR URETHRAL VALVE* Q 64.20 *

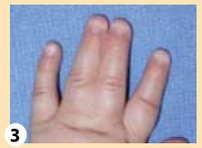
Distended bladder even after passing urine.

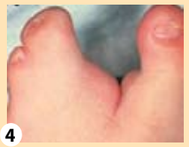
6 LIMBS (UPPER & LOWER)

1. Absence of a whole or part of upper limb
2. Absence of a whole or part of lower limb
3. Fused digits
4. Absence of digits or split hand/foot
5. Extra digits
6. Club foot


1 *LIMB REDUCTION DEFECT UPPER* Q71


2 *LIMB REDUCTION DEFECT LOWER* Q72


3 *SYNDACTYLY* Q 70


4 *ECTRODACTYLY* Q 72.7



4 *POLYDACTYLY* Q 69

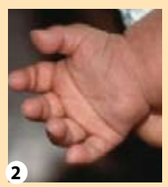


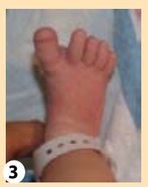
5 *CLUB FOOT-TALIPES EQUINOVARUS* Q 66.0

7 CHROMOSOMAL - DOWN SYNDROME

1. Face: Upward slanting eyes, fold on the inner corner of the eye (epicanthal), flat nose, small ear, small mouth, excess skin at the nape of neck
2. Palm: Single crease
3. Foot: Increased gap between 1st and 2nd toe


1 *DOWN SYNDROME* Q90


2


3

* *Need uraent referral*

→| *If any of the above identified, record findings in RCH register and RBSK birth defect recording format along with MCTS details.*

NATIONAL HEALTH MISSION
राष्ट्रीय स्वास्थ्य मिशन

Ministry of Health & Family Welfare
Government of India

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