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AMOGS
The Association of Maharashtra Obstetric & Gynaecological Societies

# ॥ fetopanishad ॥

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## THE FETAL BULLETIN

# THE PERINATOLOGY ROOM

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> Childbirth is more admirable than conquest, more amazing than self-defense, and as courageous as either one.
>
> *- Gloria Steinem*

TOPICS PAGE NO.



# ANTENATAL STEROIDS: TIME-TESTED FETAL THERAPY

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Respiratory Distress Syndrome (RDS) is a complication of preterm birth and primary cause of early neonatal morbidity and mortality. Preterm babies between 24-34 weeks are born with premature lungs, which are in transition from canalicular to saccular to alveolar stage of lung development.

Antenatal Corticosteroids exogenously administered cross placenta and accelerates the fetal lung maturity by its physiological effects involving increased surfactant formation and clearance of fluids after birth and cytostructural changes like narrowing of saccular septa, mesenchymal condensation and cyto-differentiation. (Fig. 1)

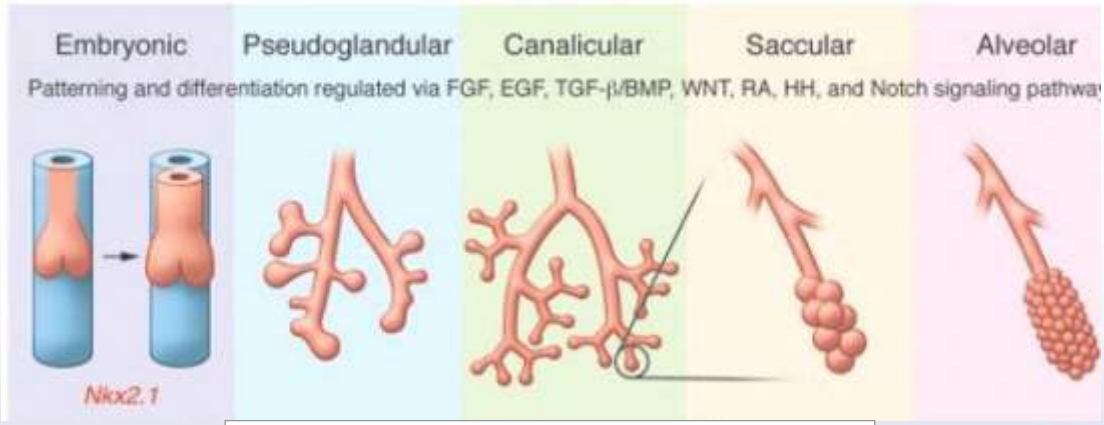



Fig 1. Depicting various stages of Lung Development

A largest systematic review published in Cochrane database in 2017 included 30 studies across the globe with total of 7774 women and 8158 infants distributed between 24-37 weeks of gestation.The review concluded statistically significant improvement in reducing the neonatal morbidity and improving the course of hospital stay (Table 1.)

TABLE 1. **Evidence of beneficial effects of antenatal steroids**

| BENEFITS | RR, 95% CI |
|---|---|
| Perinatal death | 0.72, (0.58 to 0.89) |
| Neonatal mortality | 0.69, (0.59 to 0.81) |
| RDS | 0.66, (0.56 to 0.77) |
| Moderate/severe RDS | 0.59, (0.38 to 0.91) |
| Systemic infections (first 48 hours HOL) | 0.60, (0.41 to 0.88) |

| BENEFITS | RR, 95% CI |
|---|---|
| IVH | 0.55, (0.40 to 0.76) |
| NEC | 0.50, (0.32 to 0.78) |
| Mech. ventilation | 0.68, (0.56 to 0.84) |

Devender Roberts, Julie Brown, Nancy Medley, Stuart R Dalziel. Antenatal corticosteroids for accelerating fetal lung maturation for women at risk of preterm births – SYSTEMATIC REVIEW. Cochrane Database Syst Rev. 2017 Mar 21;3(3): CD004454.

According to the evidence, corticosteroids are not beneficial in reducing chronic lung disease, neuro - developmental delay and death in childhood and adulthood. No adverse effect on educational achievements during childhood. However, statistically non-significant increase in the Insulin resistance during adulthood has been observed in a few trials.

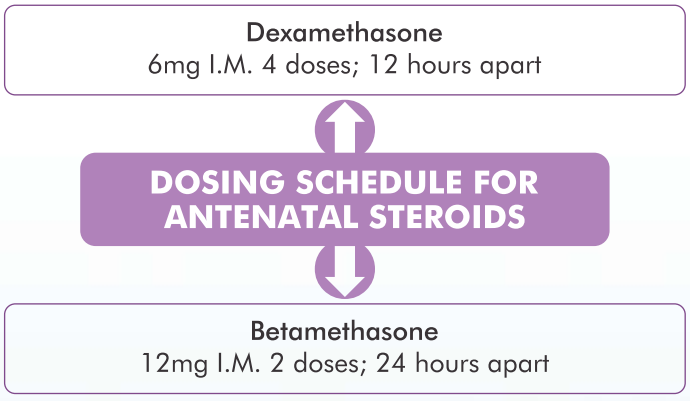
The ACOG committee opinion published in August 2017 laid down the guidelines in order to standardize the use of antenatal steroids. The guideline is summarized as follows (Table 2)

Table 2. **ACOG committee opinion**

| VARIOUS CLINICAL SETTINGS | RECOMMENDATION |
|---|---|
| Risk of preterm birth within 7 days (Between 24-33+6 weeks) | Single course |
| Periviability issues<br>1) 23 0/7 TO 23 6/7 Gestation<br>2) 22 0/7 to 22 6/7 Gestation | May consider (based on NICHD research)<br>May be considered based on family's decision regarding resuscitation |
| Preterm premature rupture of Membrane | Single course recommended |
| Multiple Gestation (24 0/7 to 33 7/6 weeks gestation) | Irrespective of the fetal number, a single course is recommended |
| Late preterm births | Single course recommended, if not received a prior complete course (based on recent evidence: ALPS Trial) |
| Serial courses | Not recommended<br>(evidence suggests increased incidence of SGA babies and maternal infection and suppression of HPA axis) |
| Single rescue course | Recommended for women <34; 0/7 weeks gestation at risk of preterm birth within 7 days with prior course administered 14 days before. |

*ACOG committee opinion, Antenatal steroid therapy. Vol 130, no.2 August, 2017

The difference between the two most commonly used steroids is summarized in the flowchart below.




| Betamethasone | Dexamethasone |
|---|---|
| Not stable at high temperature | Stable at high temperature |
| Immunosuppressive effect - weak | Immunosuppressive effect - weak |
| Longer half life and Expensive | Shorter half life & less expensive |
| Larger volume of distribution | Smaller volume of distribution |

Fig 2. Choice of steroids for fetal therapy

**Operational guidelines - Govt. of India**

- Recommend use of Dexamethasone
- Prereferral dose of steroids for mothers with high-risk of preterm delivery between 24-34 weeks
- Empowering auxiallry nurse midwife
- All elective LSCS should be done at or after 39 weeks of gestation in uncomplicated cases

Fig 3. Operational Guidelines by GOI

Over the last decade, enormous evidence for antenatal steroids has established it as the most important fetal therapy. Currently, the need of the hour is a good nationwide quality improvement strategy to optimise appropriate and a timely administration to prevent neonatal morbidity and mortality, because prevention is always better than cure.

# REDUCING NEONATAL SEPSIS: ROLE OF AN OBSTETRICIAN

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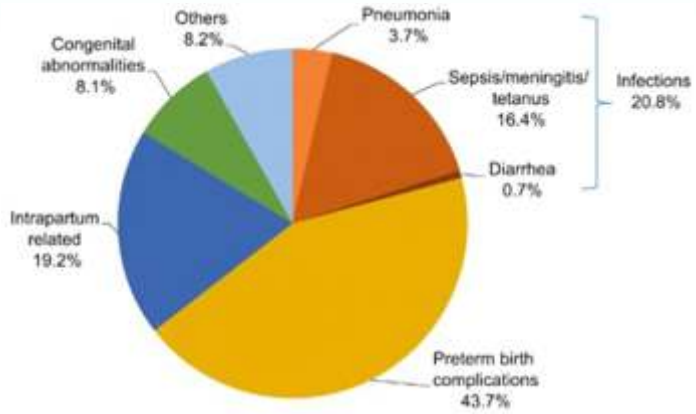
Globally, of the three million annual neonatal sepsis cases, India has the highest incidence of clinical sepsis (17,000/100,000)[1]. The role of an obstetrician is indeed important in decreasing neonatal morbidity, since proper antenatal care can decrease the risk of maternal infection and other factors leading to prematurity and neonatal complications. The mother and newborn form a dyad, and the well-being of the mother at the time of birth, directly determines the well-being of the newborn at birth. Infection in a newborn is closely linked to an infection in a mother. Maternal infections are usually associated with Early Onset Neonatal Sepsis defined as onset of infection within 72 hours of birth.



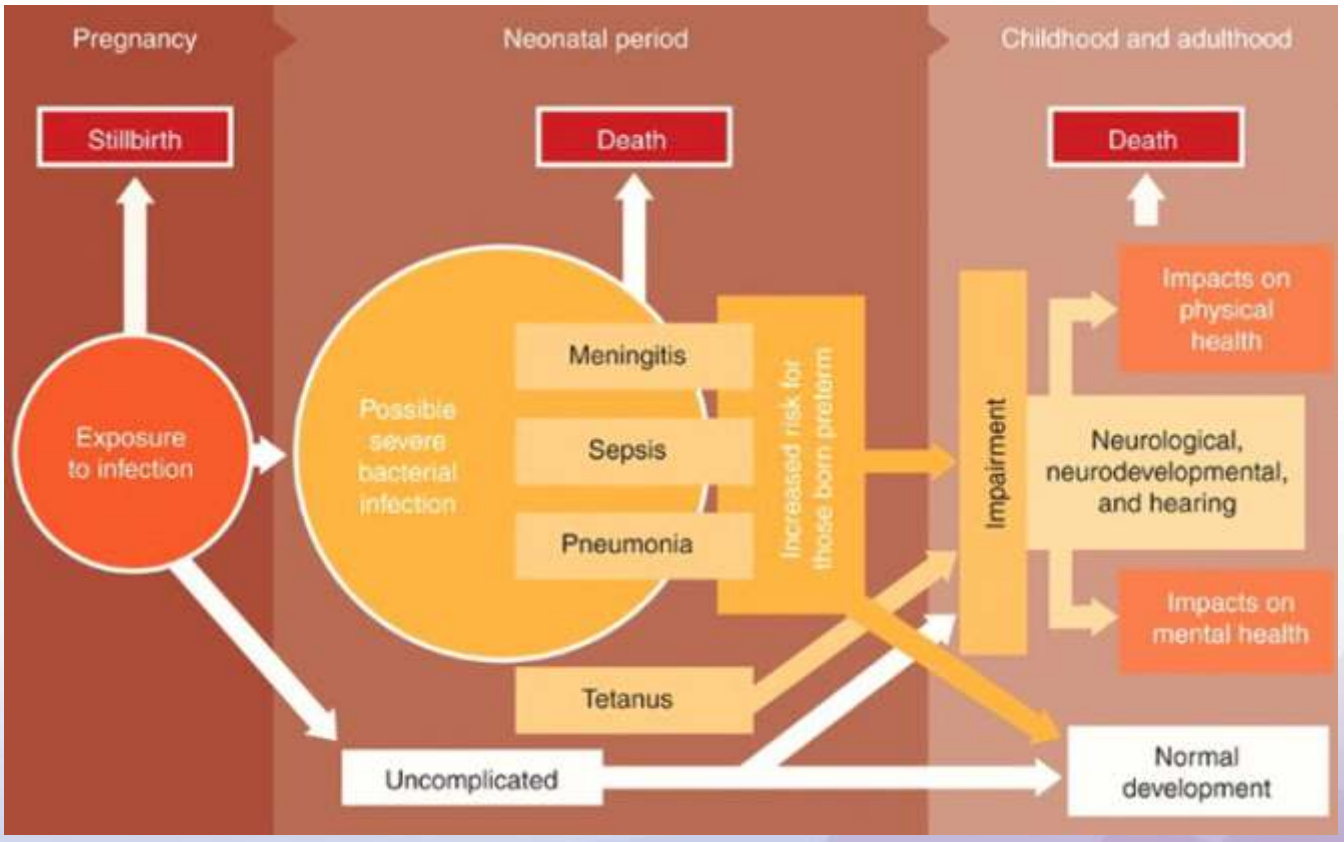

## Preconception care

Risk assessment, screening, and treatment for specific infections like TORCH, HIV, SYPHILIS and PERIODONTAL disease, should be a component of preconception care because there is convincing evidence that treatment of these infections before pregnancy, prevent neonatal infections[2].

## Antenatal and intrapartum care

Hand Hygiene: Washing hands and using High-Level Disinfected (HLD) gloves have been found to decrease the introduction of contamination through the vaginal canal and into the uterus.

Unhygienic vaginal examination poses the risk of contaminating the amniotic fluid which when ingested or aspirated by the fetus, can lead to EONS in postnatal life.



**Partograph:** Partograph guidelines limit vaginal exams to one in every four hours, thus reducing opportunities to introduce organisms into the vagina or cervix. The partograph also reduces sepsis in the newborn by reducing prolonged labour, assisted vaginal delivery and emergency caesarean sections - three factors that increase the risk of sepsis.[3]

**Clean delivery practices:** Using a plastic sheet during delivery, a boiled blade to cut the cord, a boiled thread to tie the cord and an antiseptic to clean the umbilicus, were each significantly associated with relative reductions in mortality due to sepsis.

**Respectful maternity care:** Disrespectful and undignified care is prevalent in many facility settings globally, particularly for underprivileged populations, and this not only violates their human rights but is also a significant barrier to accessing intrapartum care services.[4]

## Neonatal Sepsis

### Major Risk Factors

- Ruptured membranes>24 hrs.
- Maternal Fever 100.4°F (38°C)
- Chorionamnionitis
- Sustained fetal heart rate>160/min
- Multiple obstetric procedures

### Minor Risk Factors

- Ruptured membranes>12 hrs.
- Foul smelling liquor
- Maternal Fever>99.5℉ (37.5℃)
- Low APGAR <5 at 1 min, <7 at 5 min
- Prematurity
- Multiple gestation

**Presence of 1 major or 2 minor risk factors -> High-risk of Sepsis**

### Intrapartum practices that reduce the risk of sepsis

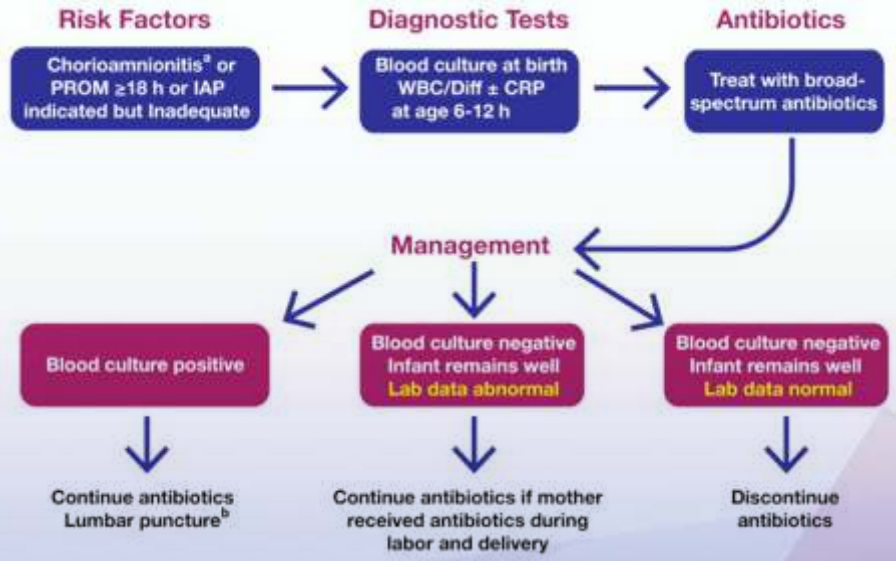
- Hand hygiene, HLD gloves
- The 'Six Cleans'
- Minimization of vaginal examinations
- Use of partograph
- Prompt diagnosis and treatment of prolonged labour
- Prevention, prompt diagnosis and treatment of chorioamnionitis

### Intrapartum practices that must be avoided

- Routine vaginal examinations at shift change
- Multiple vaginal examinations
- Vaginal examinations after rupture of membranes
- Shaving of the genital area
- Enemas

1. **Intrapartum Antibiotic Prophylaxis:** This approach has been found to be effective in reducing neonatal sepsis not only due to Group B streptococcus, but also Non-Group B Streptococcus in mothers with risk factors. It was found to be significant and went on to be identified amongst the independent risk factors for EONS such as clinical chorioamnionitis, repeated per vaginal examinations (3 clean and 1 unclean examination), male sex, birth weight <1500gms, gestation less than 30 weeks, lack of intrapartum antibiotic prophylaxis.[5]

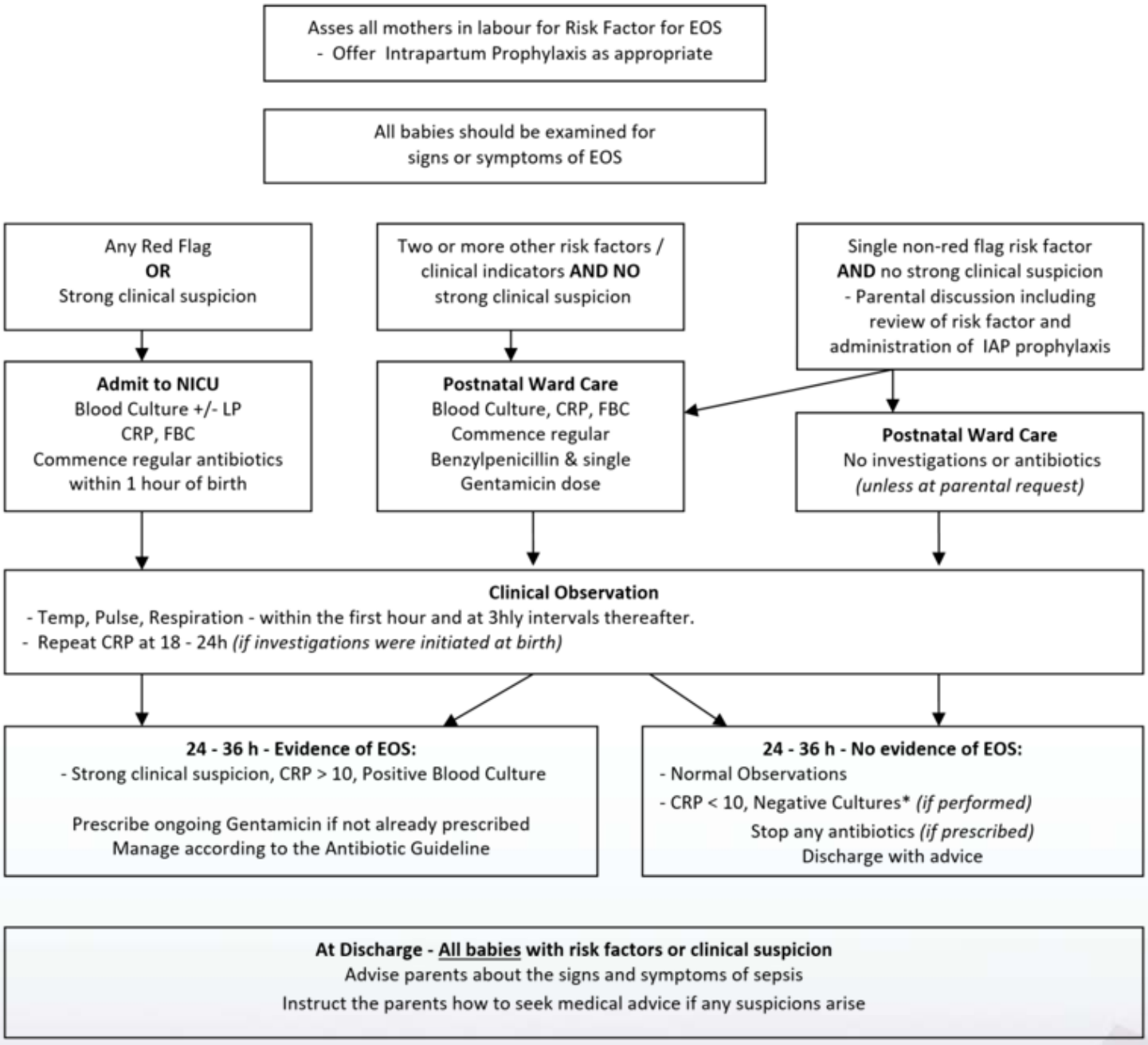
   Chorioamnionitis is now better defined by the term – Triple I, which includes Intrauterine Inflammation or Infection at birth.[6] This is defined by:
   - Fetal tachycardia
   - Maternal leukocytosis (>15000 cells in the absence of corticosteroids)
   - Purulent fluid from the cervical OS
   - Biochemical or microbiologic amniotic fluid changes consistent with infection
   - Fever (>39.0°C/102°F)

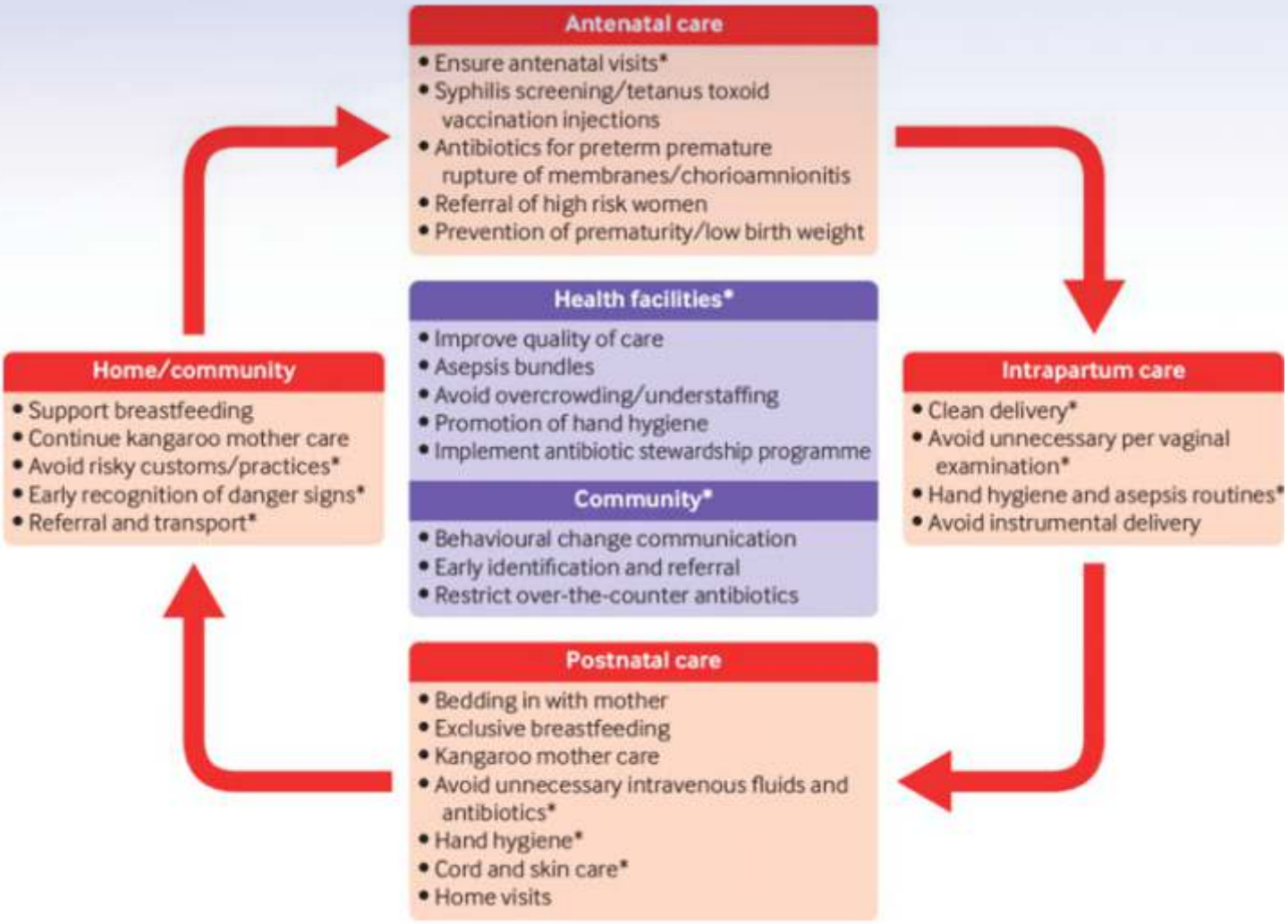


2. **Maternal immunization and antenatal steroids:** Immunization of expectant mothers is an important population strategy for providing the unborn fetus and the neonate with protective antibodies. Besides immunization with tetanus toxoid and influenza, maternal immunization with Streptococcus agalactiae type III conjugate vaccine, has been successfully used for reducing the incidence of sepsis in neonates.
   Antenatal steroids are strongly recommended because their use results in a reduction in systemic infections in the first 48 hours of life, compared to not using them in the antenatal period.[7]

3. **LaQshya initiative:** Recently, the Union Ministry of Health and Family Welfare has launched "LaQshya" (Labour room Quality Improvement Initiative) to improve the quality of care in the labour room and maternity operation theatres in public health facilities, intended to bring down the rates of neonatal sepsis.

4. **Immediate Postnatal/Newborn care:** Initiation of breastfeeding in the delivery room itself in asymptomatic neonate of >34 weeks gestational age, checking for retracted nipples, Cord Care and Eye Care of the newborn at the time of delivery, can reduce newborn infection. Early initiation of exclusive breastfeeding, is a natural way to provide antibodies that will protect the newborn from both, intestinal and respiratory tract infections. Pneumonia and diarrhea remain major causes of neonatal death, and both are reduced with exclusive breastfeeding.[8]






References :

1) Fleischmann-Struzek C, Goldfarb DM, SchlattmannP, Schlapback LJ, Reinhart K, Kissoon N. The global burden of neonatal sepsis: a systemic review. Lancet Respir Med. 2018;6(3):223-230.

2) Lassi, Z.S., Imam, A.M., Dean, S.V. et al. Preconception care: preventing and treating infections. Reprod Health 11, S4 (2014). https://doi.org/10.1186/1742-4755-11-S3-S4.

3) Haws RA, Yakoob MY, Soomro T, Menezes EV, Darmstadt GL and Bhutta ZA. 2009. Reducing stillbirths: Screening and monitoring during pregnancy and labour. BMC Pregnancy Childbirth 9(Suppl 1): S5.

4) Bohren MA, Hunter EC, Munthe-Kaas HM, Souza JP, Vogel JP, Gülmezoglu AM. Facilitators and barriers to facility-based delivery in low and middle-income countries: a qualitative evidence synthesis. Reprod Health. 2014;11

5) Dutta S, Reddy R, Sheikh S, Kalra J, Ray P, et al. (2010) Intrapartum antibiotics and risk factors for early onset sepsis. Arch Dis Child Fetal Neonatal Ed 95: F99-103.

6) Nelson textbook of Pediatrics e21, vol. 1; pg 998-999.

7) 2. Roberts D, Dalziel S (2006) Antenatal corticosteroids for accelerating fetal lung maturation for women at risk of preterm birth. Cochrane Database SystRev : CD004454.

8) Black RE, Cousens S, Johnson HL, Lawn JE, Rudan I, Bassani DG, Jha P, Campbell H, Walker CF, Cibulskis R, Eisele T, Liu L, Mathers C; Child Health Epidemiology Reference Group of WHO and UNICEF. 2010. Global, regional, and national causes of child mortality in 2008: A systematic analysis. Lancet 375(9730): 1969–1987.

# MGSO4 FOR NEUROPROTECTION

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## Introduction

Increasing prevalence of preterm birth, that is birth before 37 weeks of gestation, is the single most important determinant of adverse infant outcomes, in terms of survival and quality of life. Indications for preterm deliveries are both, maternal and fetal and in India the incidence is approximately 7-9% of births.While the survival of infants born preterm has improved, the prevalence of cerebral palsy has risen with profound emotional, medical, and financial consequences.This article discusses the benefits of use of antenatal MgSO4 administration for fetal neuroprotection.

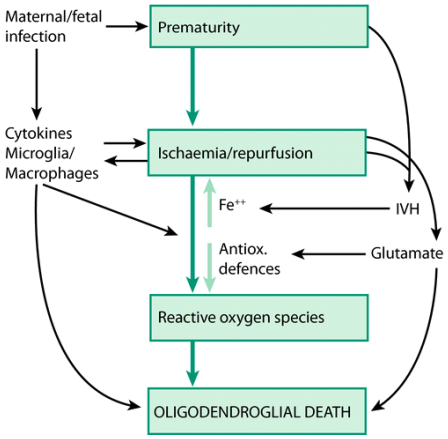
## Discussion

The incidence of cerebral palsy decreases significantly with increasing gestational age: 14.6% at 22–27 weeks of gestation, 6.2% at 28–31 weeks, 0.7% at 32–37 weeks and 0.1% in term infants. Infant mortality and morbidity from preterm birth can be reduced through interventions delivered to the mother before or during pregnancy, and to the preterm infant after birth. The most beneficial set of maternal interventions are those that are aimed at improving outcomes for preterm infants when preterm birth is inevitable (e.g. antenatal corticosteroids, magnesium sulfate and antibiotic prophylaxis).

## Definitions

| TERM | GESTATIONAL AGE |
|---|---|
| Moderate to late Preterm | 32-37 weeks |
| Very Preterm | 28-32 weeks |
| Extremely Preterm | <28 weeks |
| Extreme Preterm | |

## Risks for preterm




The ensuing cerebral lesions are strongly associated later with cerebral palsy and neurobehavioral developmental disorders. The mechanisms that lead to such damage are unclear, but vascular instability and oxygenation of the immature brain appear to play a part. Although no single neuroprotective intervention is known to prevent preterm brain injury, neuroprotective strategies should be adopted to improve the outcomes.

## Intervention

One such intervention is antenatal administration of Magnesium Sulfate (MgSO4) in women at risk of preterm birth.

Magnesium Sulfate for Prevention of Eclampsia (MAGPi) trial had already proven the safety of use of MgSO4 in pregnant women. Various studies like Australasian Collaborative Trial of Magnesium Sulphate (ACTOMgSO4) and beneficial effects of Antenatal Magnesium Sulfate (BeAM) also have proven the reduced incidence of poor neonatal outcomes like cerebral palsy with MgSO4 use.The benefit remained similar regardless of gestational age, cause of prematurity, and total dose received in various studies.

Antenatal Magnesium Sulfate is now recommended by the WHO and many pediatric and obstetrical societies, too.

## Potential mechanism of action of MgSO4

One of many ways magnesium acts, is by contributing to glycolysis and ATP production, as well as functions as a cell membrane stabilizer.

## Effects of MgSO4

Magnesium affects several pathways potentially involved in preterm brain injury. Magnesium sulfate given prior to delivery can possibly reduce the risk as it is known to stabilize the vasculature and reduce hypoxic effects by mitigating cytokine or excitatory amino acid damage. Magnesium also has anti-inflammatory properties.

## Dose schedules

Magnesium sulfate for neuroprotection should only be given if preterm birth is likely within the next 24 hours.

1. IV 4 g over 20 minutes, then 1 g/hour until delivery or for 24 hours, whichever came first
2. IV 4 g over 10–15 minutes, followed by either IV 1 g/hour for 24 hours, or by IM 5 g every 4 hours for 24 hours
3. Single dose of IV 4 g over 30 minutes

There was insufficient evidence to recommend one specific dosing regimen over others. This dose applies to women carrying either singleton or multiple pregnancies. There is a need for further research to establish whether repeated treatment with Magnesium Sulfate for neuroprotection is appropriate (i.e. in the event that delivery does not occur).

## Conclusion

MgSO4 probably has a modest neuroprotective effect which is greater the earlier the gestational age of the infant at delivery and should be preferred for use in preterm deliveries less than 30-32 weeks of gestation. When administered at an appropriate dose and with proper monitoring, there is no evidence of harm to the fetus, neonate or mother.

# PLACENTAL TRANSFUSION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The theory of placental transfusion dates back from the era of Darwin in the 18th century. Placental transfusion occurs in all mammals, exception being only when we humans clamp the cord earlier. After birth when the uterus continues to contract, blood also continues to flow via the umbilical vessels to the newborn for a few minutes facilitating a transfusion of additional 80-100ml of blood.

Historically, this theory of more placental transfusion has changed vividly with the latest acceptance of delayed cord clamping universally. The definition of delayed cord clamping hasn't yet reached a uniform consensus. But based on evidence from systematic reviews, delayed cord clamping could be defined as greater than one minute or when cord pulsation has ceased.

In term infants, more placental transfusion has been shown to reduce the chances of anemia with increased ferritin levels and total body iron stores in infancy. No significant difference in clinical jaundice and symptomatic polycythemia has been found. In preterms, more placental transfusion increases cerebral oxygenation with avoiding the disruption in cerebral blood flow autoregulation, therefore lower risk of all grades of intraventricular hemorrhage. Also, in preterms with delayed placental perfusion, there has been a lesser need for transfusion, better circulatory stability and lower risk of necrotizing enterocolitis.

Placental transfusion can occur through cord milking with an intact cord attached to the placenta facilitating increasing placental blood flow to the brain and lungs and also via Cut-umbilical cord milking performed by clamping away from the fetus and retaining a long segment of the umbilical cord that can be milked by the neonatologist even during resuscitation.

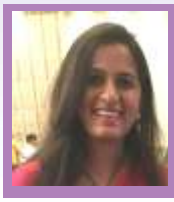
ACOG, RCOG and WHO, all recommend a delay in umbilical cord clamping in vigorous term and preterm infants for at least 30-60 seconds after birth.

Also advocates the documentation of timing of cord clamping routinely with cord milking being a good alternative.

Summarizing the benefits of delayed placental transfusion as follows:

| Immediate benefits | | | Long-term benefits | |
|---|---|---|---|---|
| **Preterm/low-birth-weight infants** | **Full-term infants** | **Mothers** | **Preterm/low-birth-weight infants** | **Full-term infants** |
| Decreases risk of:<br>• intraventricular haemorrhage<br>• necrotizing enterocolitis<br>• late-onset sepsis | Provides adequate blood volume and birth iron stores | No effect on maternal bleeding or length of the third stage of labour | Increases haemoglobin at 10 weeks of age | Improves haematological status (haemoglobin and haematocrit) at 2–4 months of age |
| Decreases need for:<br>• blood transfusions for anaemia or low blood pressure<br>• surfactant<br>• mechanical ventilation<br>Increases:<br>• haematocrit<br>• haemoglobin<br>• blood pressure<br>• cerebral oxygenation<br>• red blood cell flow | Increases:<br>• haematocrit<br>• haemoglobin | Indication from "cord drainage" trials that less blood-filled placenta shortens the third stage of labour and decreases the incidence of retained placenta | May be a benefit to neurodevelopmental outcomes in male infants | Improves iron status up to 6 months of age |

# RETINOPATHY OF PREMATURITY - OBSTETRICIANS' ROLE



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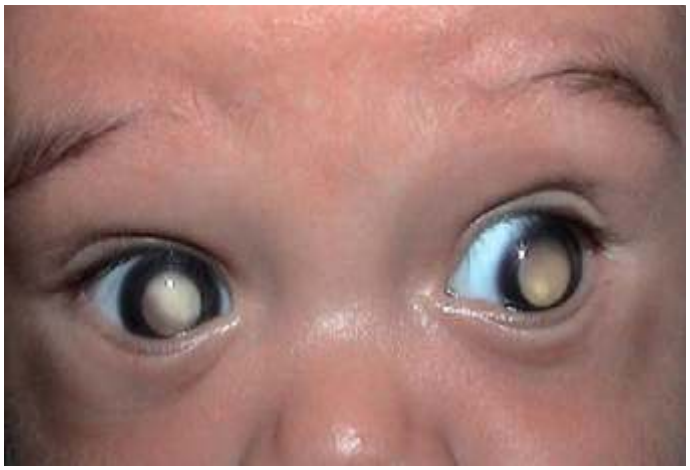
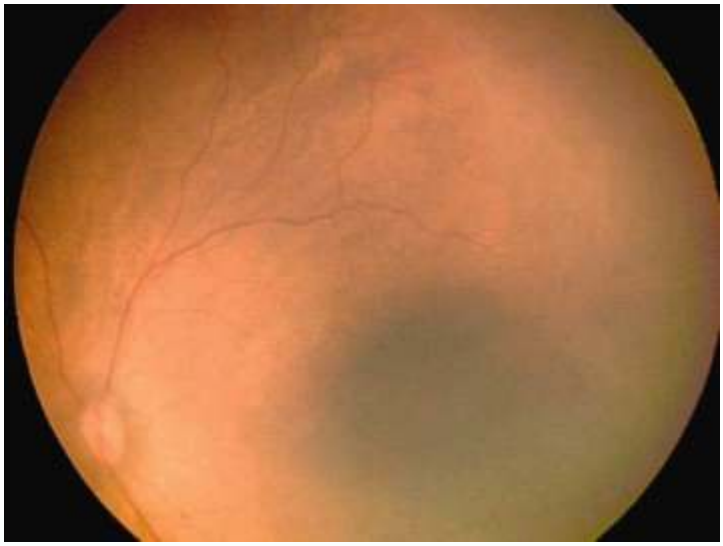
## Keeping an eye on the preemie's eye

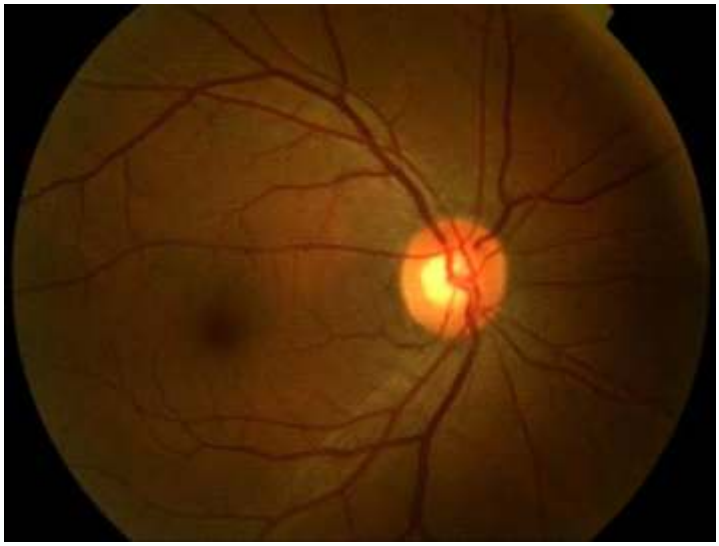


You see a child like this, white reflex in both eyes, preterm, low birth weight, NICU stay with O2 therapy given – and you know a disaster has occurred. Retinopathy of prematurity is one of the most important causes of "preventable" blindness, more so in a country like ours where preemies account for around 10% of all the newborns.

As obstetricians, the responsibility of awareness and prevention falls mightily on your shoulders - the OBG fraternity often being the first medical point of contact of the parents, the first doctor having taken care of their unborn child.

Let me take you through the journey of retinal vascular development, and briefly on how ROP actually develops.





The first image is that of a 30 weeker, where areas of peripheral retina have not been vascularised, retinal arteries ending prematurely. The image on the right is that of a fully developed, vascularised retina.

It is usually at term that the retinal vascular development nears completion. Avascular retina in preterms causes hypoxia, which in turn, leads to new vessel formation causing complications like retinal detachment in these children.

## Where does an obstetrician's part come in to play here?

BEFORE BIRTH:

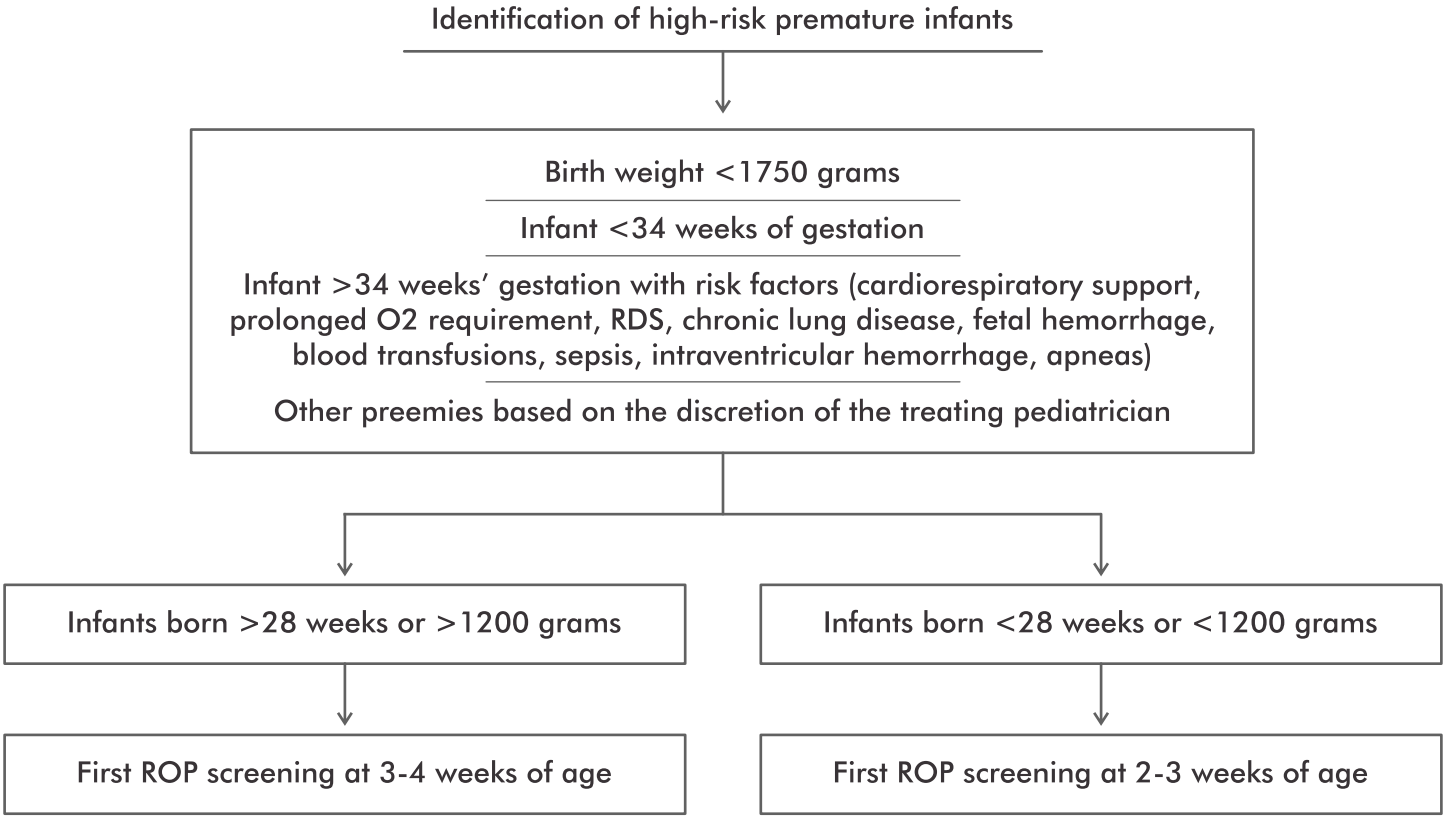
- EVERYDAY matters in the progress of retinal development. So, try pushing delivery of the child as much - weighing risk and benefit ratio. A study on South Asian eyes revealed 91.6% babies at <25 weeks of gestation, needed intervention in the form of surgery or laser as against 41.7% at >29 weeks of gestation
- Sensitize parents to the importance of screening and early treatment, as visual milestones are usually ignored early in life, and later it is too late for these children

AFTER BIRTH:

- Refer babies at risk, following the guidelines
- Try to sensitize neonatologists to good ICU practices to reduce the load of preemies, that go into stages of ROP needing treatment. When immediate neonatal care is not available, consider judicious use of oxygen in obstetric settings
- Encourage patients to go for regular eye screening as morbidity with ROP does not end at just the first few months. Long-term complications include high myopia, nystagmus, cataract, glaucoma and late retinal detachments

The prevention and treatment require a multidisciplinary approach and together we can reduce the suffering of these families bogged down by the adversary - that is retinopathy of prematurity.

## ALGORITHM FOR REFERRAL OF ROP BABIES



# FAQs ON NEWBORN SCREENING (NBS)

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## 1. When is NBS ideally carried out?

Screening in neonates should ideally be undertaken between **3-28 days of birth** (in neonatal period), to detect common and treatable endocrine, Inborn Errors of Metabolism (IEM), hemoglobinopathies and issues such as hearing loss or congenital heart defect.

## 2. Are Metabolic Disorders common in India to warrant screening?

In India, the incidence of Inborn Errors of Metabolism, is far more common compared to the West because of factors such as consanguinity (marriages in relationship) or endogamy (marriages in small communities). The combined incidence of IEMs in India is as high as **1:2000**.

## 3. NBS should be considered for which disorders? Are there any guidelines?

National Neonatal Forum (NNF) guidelines for Newborn Screening classifies patients into 3 categories:

**Category A**: Ideally to be considered in **all newborns**. This category includes screening for3 most common conditions - Congenital Hypothyroidism, Congenital Adrenal Hyperplasia and G6PD deficiency and if possible, it is also advised to include Galactosemia, Biotinidase deficiency and Phenylketonuria along with newborn hearing screening.

**Category B**: Includes **high-risk neonates**, critically-ill neonates, newborns with symptoms and signs suggestive of Inborn Errors of Metabolism, previous siblings with unexplained mental retardation, seizure disorder, delayed development, unexplained sibling deaths and consanguinity. In these group of patients apart from screening for common disorders, Tandem Mass Spectrometry (TMS) is advised to look for Organic acidurias, Aminoacidopathies, Fatty acid oxidation defects and Hemoglobinopathies.

**Category C**: Includes patients in **resource available settings**. Advanced Newborn Screening should be considered in such group of patients.

## 4. In which newborn cases should Inborn Errors of Metabolism be strongly suspected?

**All critically-ill neonates requiring NICU admission**, newborns with signs and symptoms suggestive of IEM (altered sensorium, lethargy, irritability, poor feeding, seizures, diarrhea, any intractable presentation etc.), or a **contributory family history** such as previous sibling with mental retardation, developmental delay, neurological issues or unexplained sibling deaths. NBS must also be considered in cases with suspected sepsis or hypoxic ischemic encephalopathy, as many of these are a result of an underlying IEM.

## 5. When should the samples be collected for NBS?

Ideally, samples should be collected between **48-72 hours after initiating feeds** in term babies, but may be collected just before discharge from the nursing home. For preterm babies screening should be initiated when they reach term gestational age, but screening can be done anytime in critically-ill neonates before initiating emergency treatment.

## 6. How are the samples collected?

Blood should be collected using the heel prick method where the sample needs to be taken with utmost caution. About **4-5 blood spots** are required for processing the test. This is a simple procedure and doctors or nursing staff can be easily trained for the same. Alternatively, 1ml blood may be collected in a heparinized vacutainer or tube.

## 7. What is the impact of Newborn Screening?

The impact is most evident for Congenital Hypothyroidism where initiating early treatment can prevent intellectual disability and growth delays. Similarly, treatment is simple and has a fairly good outcome for conditions such as Congenital Adrenal Hyperplasia, Galactosemia, G6PD deficiency, Biotinidase deficiency etc. But for conditions like amino acid and organic acid disorders, the treatments are more cumbersome and involve life-long medication and dietary restrictions. But the outcome in certain conditions is excellent and prevents neuro-disability, if detected on time and managed well. Also screening for hearing loss and cardiac disorders prevents delay in initiating appropriate rehabilitation or treatment.

## 8. What is the principle of treating an IEM?

The medical management helps in removal of toxic chemicals from the body and provides the deficient nutrient/molecule.

The dietary management prevents the accumulation of harmful substances by excluding the offending nutrient. This can be achieved by using special medical food that can help in correcting metabolic abnormalities and elimination of toxic metabolites from the body.

## 9. How are these conditions managed in long-term?

Multidisciplinary team approach and coordination is needed between the Pediatrician, Metabolic Geneticist and the Metabolic Dietitian for better patient outcomes. An appropriate intervention based on the biochemical parameters is necessary at every stage, to ensure age appropriate growth and development and to improve quality of life in these patients who are suffering from IEM.

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