Prenatal screening and Prevention of Birth Defects

Dr Bharti Sharma

Birth defect burden

- Worldwide 7.9 million births occur annually with serious birth defects and 94% of these births occur in middle and low-income countries.
- Birth defects account for 7% of all neonatal mortality and 3.3 million under-five deaths.
- In India birth defects prevalence varies from 61 to 69.9/1000 live births.
- Major birth defects include **congenital heart defects**, **neural tube defects** and **Down syndrome**, **hemoglobinopathies,** and **glucose-6-phosphate dehydrogenase deficiency**
- **It has been estimated that 70% of birth defects are preventable.**

World Health Organization. Management of birth defects and hemoglobin disorders: Report of a Joint WHO-March of Dimes meeting. Geneva, Switzerland, Geneva: WHO; 2006.

Causes of Birth defects

Genetic 25%	Environmental - 15%	Complex (Multifactorial)-60%
• Down syndrome • Mendelian single-gene defects • Age • Consanguineous marriages	• Infections – rubella • Maternal diseases – DM, High fever • Teratogenic drugs, Alcohol Smoking Environmental pollutants	• Gene-environmental interaction eg.Isolated neural-tube defects, orofacial clefts

Types of birth defects

- **Lethal defects:** cause stillbirth or infant death or pregnancies are terminated after the prenatal diagnosis. eg- Anencephaly, Renal agenesis, or Hypoplastic left heart syndrome
- **Severe defects:** without medical intervention cause handicap or death. eg- cleft lip, congenital pyloric stenosis
- **Mild defects:** require medical intervention but life expectancy is good. eg-congenital dislocation of the hip or undescended testis

Major risk factors for Indians

- Large number of unplanned pregnancies
- Poor coverage of antenatal care
- Poor maternal nutritional status
- High consanguineous marriage rate
- High carrier rate for hemoglobinopathies

Unplanned pregnancies and no antenatal care

- Unplanned pregnancies and no antenatal care straightway mean pregnancies do **not benefit from preventive strategies against birth defects.**

- **Antenatal care- NFHS-5 (2019-21)**

  Mothers who had at least 4 ANC visits -58.1 %

  Mothers who had antenatal check-up in the first trimester -70%

  Registered pregnancies for which the mother received a Mother and Child Protection (MCP) card (95.9%)

Medical condition of mother

- Exact prevalence of chronic conditions like diabetes, epilepsy, hypertension during pregnancy is not known.
- About 8% of pregnant women need permanent drug treatment due to various chronic diseases and pregnancy-induced complications.

Maternal nutritional status

- Maternal deficiencies of iodine, folic acid, and other macro and micronutrients associated with birth defects.
- Mothers who consumed iron folic acid for 100 days or more when they were pregnant- 44.1%
- According to NFHS 3, just over half (51%) of households were using salt that was adequately iodized, NFHS 4 -93% NFHS-5 94.3%
- 52.2% of pregnant & 57.2% of nonpregnant women were Anaemic
- High prevalence of nutritional deficiency -18.7% of women have a BMI below 18.5 & Obesity 24%

NFHS-5 (2019-2021)

Consanguineous marriages

- Consanguinity rates in India varies from as low as 1% to 4% in the northern region to as high as 40-50% in the southern region.
- In comparison to a non-consanguineous couple, consanguineous are more likely to have

  Early age at marriage and at first birth

  Higher number of infants born

  Higher rates of postnatal mortality

  Higher rates of congenital malformations and genetic disorder.

Parent’s carrier status of a genetic disorder

- Carrier frequencies for various genetic disorders are high among Indians.
- **Sickle cell hemoglobin** - 17% to 30% or more in the population.
- **Hb E** - found in the eastern half of the Indian sub-continent, and throughout South-East Asia, where carrier rates may exceed 60% of the population in some areas.
- **β thalassaemia** ranges from 0.3% to 15%, while that for the milder forms of α thalassemia varies from 15% to 80% (tribal population) in northeastern parts of India.

Exposures to teratogens

- According to NFHS 5- 8.9% of reproductive age group females were using any form of tobacco.
- Children of women who smoke during pregnancy are found to have multiple birth defects 1.5-2 times more than expected
- Alcohol- Fetal alcohol syndrome, usage among reproductive age group females was 1.3% (NFHS-5).
- Exposures to other categories of drugs during their first trimester vary from 55.28% for category A to 6% for category D drugs *(DRUG UTILIZATION PATTERN DURING PREGNANCY IN NORTH INDIA 2006)*
- Easy availability of drugs, intake of non-prescribed drugs, and self-medication.

Prevention

Primary	• Avoiding the cause
Secondary	• Early detection followed by effective early treatment
Tertiary	• complete recovery of congenital abnormalities by early surgical intervention without residual defects or minimal after effects.

Primary Prevention

- Right care
- Peri-conceptional folic acid
- Rubella vaccination
- Maintain a healthy weight
- Planned pregnancy
- Optimization of underlying disease- DM, Epilepsy
- Antenatal care –At least 4 antenatal visits (8 antenatal visits)
- USG 18-20 weeks to rule out anomaly (WHO)

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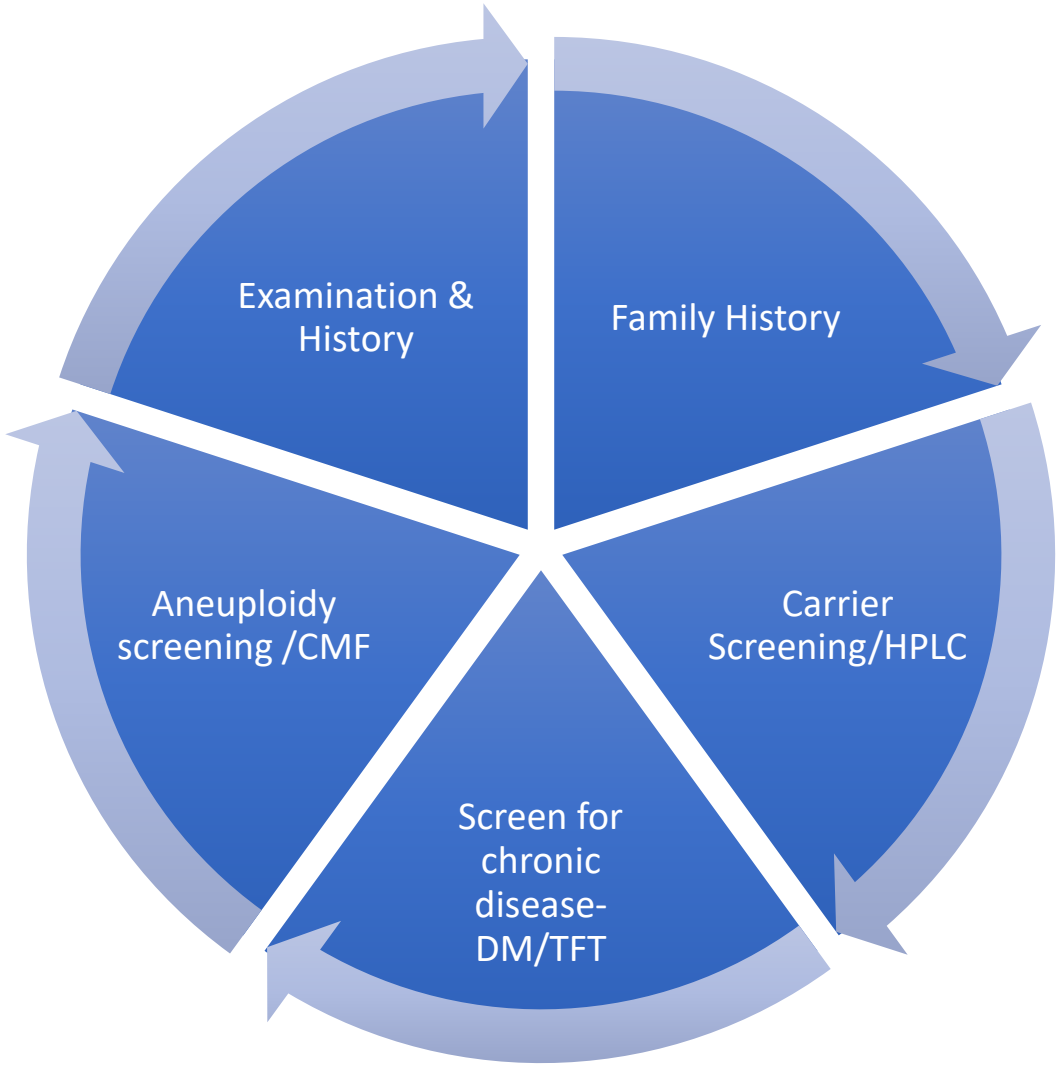
Right care


Examination & History
Family History
Carrier Screening/HPLC
Screen for chronic disease- DM/TFT
Aneuploidy screening /CMF

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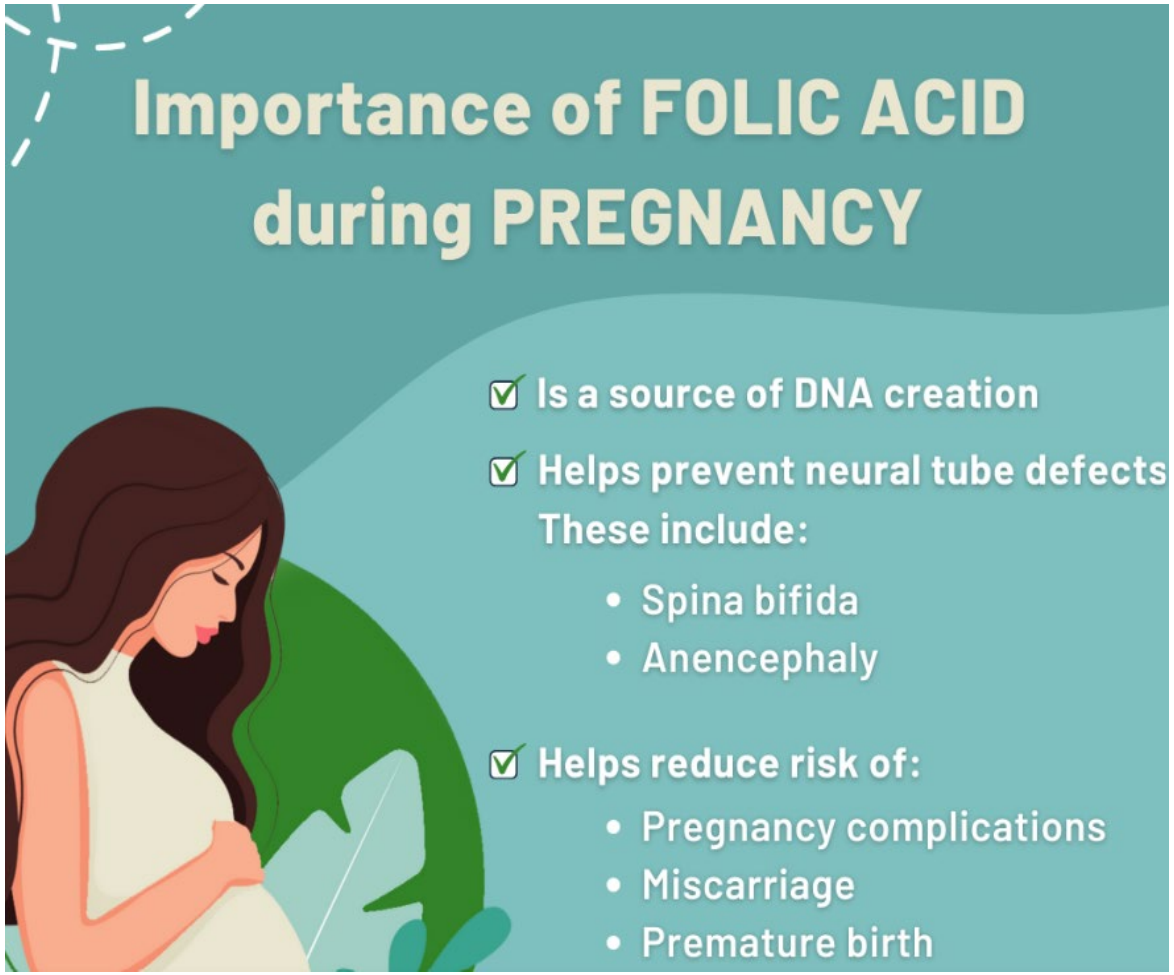
Peri-conceptional Folic acid



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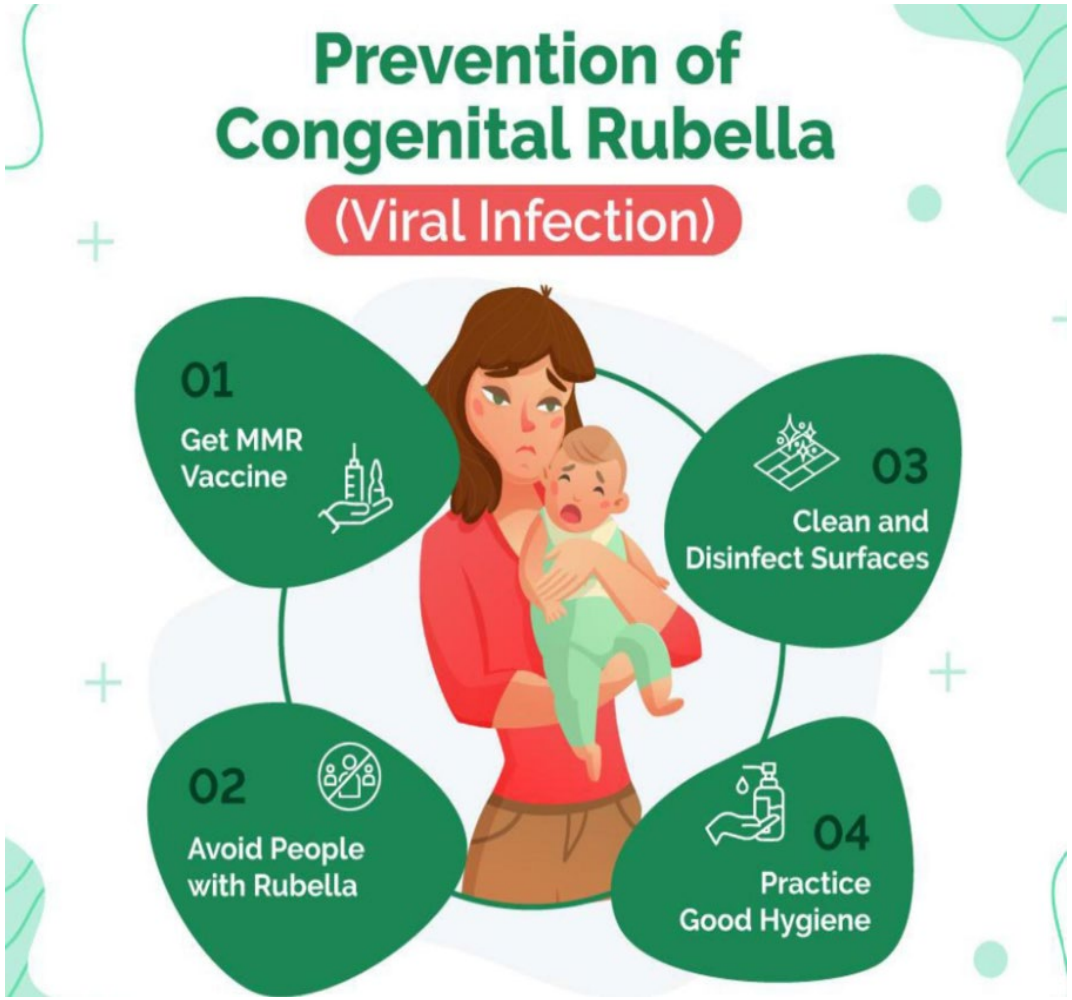
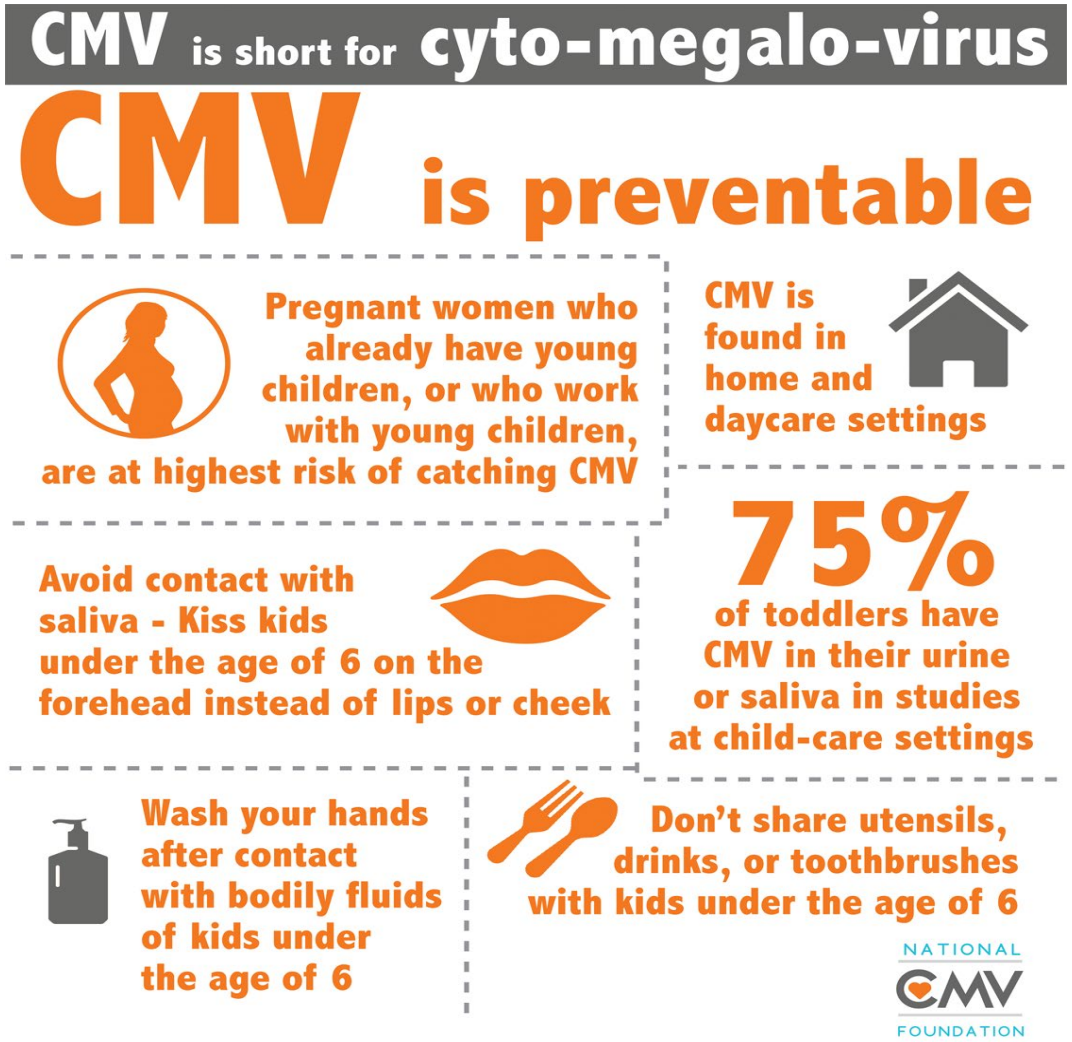
Intrauterine infections





Intrauterine infections-Toxoplasmosis

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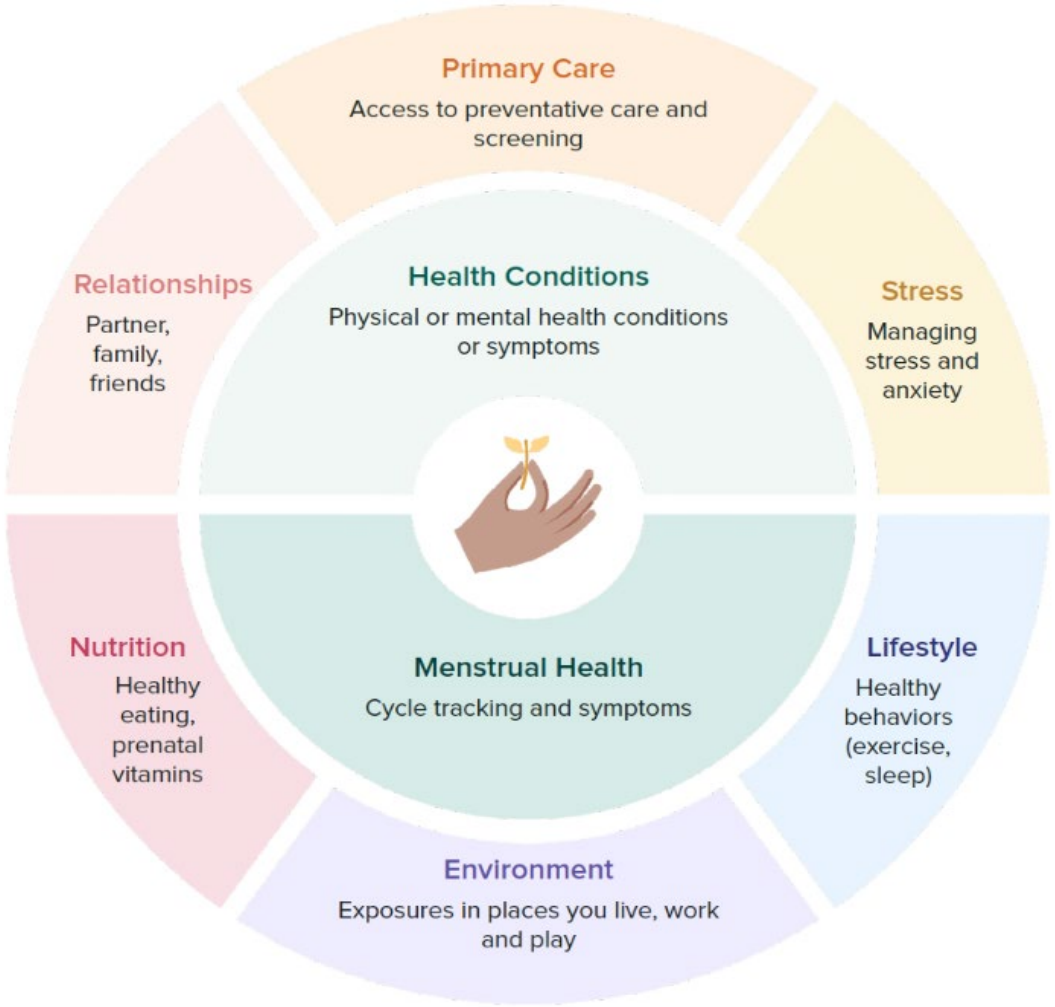
Antenatal care visits

WHO FNAC model	2016 WHO ANC model
First trimester	
Visit 1: 8-12 weeks	Contact 1: Up to 12 weeks
Second trimester	
	Contact 2:20 weeks
Visit 2: 24-26 weeks	Contact 3:26 weeks
Third Trimester	
Visit 3: 32 weeks	Contact 4:30 weeks
	Contact 5:34 weeks
Visit 4: 36-38 weeks	Contact 6:36 weeks
	Contact 7:38 weeks
	Contact 8:40 weeks
Return for delivery at 41 weeks if not given birth	

To summarize -Right care



Secondary prevention

- **Level 2 USG 18-22 weeks of gestation**

  Elective termination of pregnancy after the prenatal diagnosis of severe fetal defects was also named as secondary prevention.

- **Neonatal orthopedic screening** - early detection and treatment of deformities such as congenital dislocation of the hip based on Ortolani click and treated with different conservative methods (e.g. Pavlik pillow).

- **Patent ductus arteriosus** can be corrected by drugs immediately after birth.

Prenatal Counselling for fetus with BD

To provide comprehensive information about

- Nature and severity of congenital anomalies
- Potential outcomes
- Available treatment
- Impact on the baby's health
- Emotional support

A multidisciplinary approach (Pediatric surgery/Cardiology/genetic consultation)

Option of MTP/Genetic tests/ Autopsy

Initiatives to prevent preventable Birth defects

- **Folic acid supplementation:** Promoting folic acid intake among pregnant women, which is known to significantly reduce neural tube defects.
- **Iodization of salt:** To address iodine deficiency, which can lead to developmental issues.
- **Nutritional counseling:** Providing dietary advice to pregnant women through antenatal care visits
- **Genetic counseling and carrier screening:** Especially for families with a history of genetic disorders
- **Awareness campaigns:** Educating the public about the importance of preconception care and preventing risk factors for birth defects
- **Food fortification programs**
- **Antenatal care , Level II Ultrasound**
- **Pradhan Mantri Surakshit Matritva Abhiyan-** High Risk pregnancies

Prevention of Neural Tube Defects

Thank you Maa,

for taking the magic pill "Folic Acid"
to prevent Neural Tube Defect (NTD)

More than 70% of NTD are Preventable

- Periconceptional **Folic Acid** for all women of child-bearing age
- ANC check-up along with Level II Ultrasound

Timely Intervention

- Surgery
- Physiotherapy

RBSK
RASHTRIYA BAL SWASTHYA KARYAKRAM
FROM SURVIVAL TO HEALTHY SURVIVAL

NATIONAL HEALTH MISSION

Thank you

for making healthy choices to Prevent Birth Defects

Pre-pregnancy Planning

- Folic Acid supplementation (400 µg/day)
- Regular intake Folic Acid rich food like green leafy vegetables, pulses
- Rubella Vaccination
- Intake of fortified food

Care during Pregnancy

- Discuss your genetic and family history with your health care team
- Get your regular antenatal checkup at least four times during pregnancy
- Inform your doctor before starting or stopping any medications

Avoid Harmful Substances: alcohol/ smoking (including passive smoking) at any time during pregnancy

- Maintain healthy lifestyle
- Maintain a healthy weight
- Keep diabetes under control

Getting Family Support

- Family members to maintain positive environment at home
- Avoid any maternal stress and domestic violence

Keeping Personal Hygiene

Child Health Division (RBSK)
Ministry of Health and Family Welfare
Government of India

Questions

Q.1 Which of the following vaccines should be taken before getting pregnant because of the danger of disease or birth defects to the fetus?

A. Tetanus
B. Diphtheria
C. Rubella
D. Flu

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A. First trimester (First 3 months)
B. Second trimester
C. Last trimester
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A. First trimester (First 3 months)
B. Second trimester (3-6 months)
C. Last trimester(6-9 months)
D. All 9 months

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