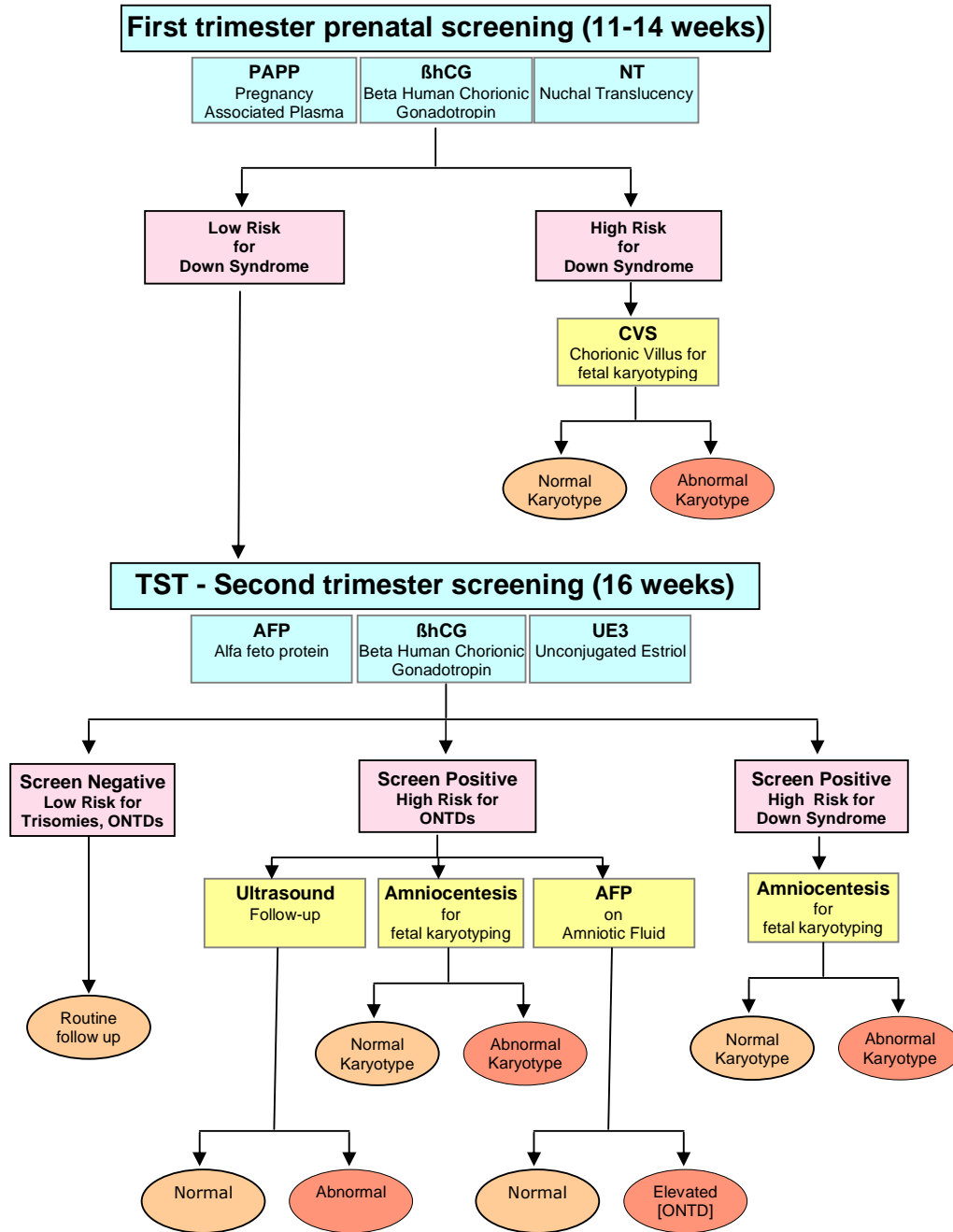




# Prenatal Screening Tests

Prenatal screening identifies a high risk group of healthy pregnant women whose risk is high enough to warrant for prenatal diagnosis. The primary objective of screening program is prevention of birth defects at a considerably lower cost and with minimal prenatal diagnostic procedures. Screening however is not an alternative to diagnosis and must be followed by prenatal diagnostic procedures to rule out or confirm a disorder. Prenatal screening is a routine test recommended for all pregnant woman as most of the mothers giving birth to Down syndrome child do not have a family history and are less than 35 years old.



## Maternal Serum First Trimester screening at 11 weeks of gestation

### free $\beta$ -hCG

Maternal serum free  $\beta$ -hCG normally decreases with gestation after 10 weeks. In trisomy 1 pregnancies, the levels are increased and the difference between these and normal pregnancies increases with advancing gestation. The detection rate using free  $\beta$ -hCG alone is 35% and in combination with maternal age the detection increases to about **45%**.

### Serum PAPP-A

Maternal serum PAPP-A normally increases with gestation. In trisomy 21 pregnancies, the levels are lower but the difference between trisomy 21 and normal pregnancies decreases with advancing gestation. The detection rate is about 40% and in combination with maternal age, the detection increases to about **50%**.

### Serum free $\beta$ -hCG and PAPP-A

Both the markers are tested and risk by taking degree of correlation between the markers. Additionally each marker showed a small but significant negative correlation with maternal weight. After combining free  $\beta$ -hCG and PAPP-A with maternal age in mathematical models, the detection rate is about **63%**.

### Serum free $\beta$ -hCG and PAPP-A and Fetal Nuchal Translucency (NT)

Nuchal translucency is the edema or thickness behind the fetal neck also called as nuchal cystic hygroma. Fetal nuchal translucency thickness increases with crown-rump length and can be measured successfully by transabdominal ultrasound examination in most of the cases. The estimated detection rate for trisomy 21 by a combination of maternal age, fetal nuchal translucency maternal Serum free  $\beta$ -hCG and PAPP-A is about **90%**.



## Maternal Serum Second Trimester screening at 16 weeks of gestation

### Serum AFP

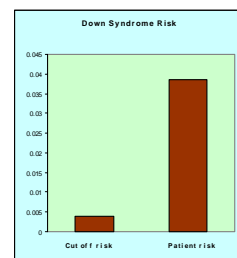
Maternal serum alpha feto protein levels indicate risk for Down syndrome when found to be decreased compared to the normal range and risk for ONTD (open neural tube defects) when found to be increased than the normal range for the gestation. The detection rate using free AFP alone is **35%** for trisomies and **90%** for ONTDs

### Serum AFP and free $\beta$ -hCG

After combining AFP and free  $\beta$ -hCG with maternal age, gestation age and maternal weight in mathematical models, the detection rate is about **55%**.

### Serum AFP, free $\beta$ -hCG and UE3 (TST)

In Triple Screen Test (TST) three serum markers are tested and the estimated detection rate for trisomy 21 is **72%**.



Apart from detecting Down Syndrome and ONTDs the screening markers also have significance in detecting other trisomies (13, 18), turner syndrome, molar pregnancies, multiple pregnancies, fetal death, gastroschisis, X-linked ichthyosis in normal and ART (artificial reproductive technology) conceptions.

## Technique and Risk Calculation

Highly sensitive "Time resolved fluorescence technique" or "Fluorimetry" is used to quantitate marker values. The values are expressed as Multiples of Median (MOM). Risk for specific disorders is calculated by clinically validated screening management software and is compared with the estimated cut off risk for the population. Accurate information like maternal age, weight, IDDM, gestation age are required for correct risk calculations. Sample required is whole blood or serum.



### References:

1. Wheeler DM et al. *Prenat Diagn* 1998; 18: 537-43
2. Spencer K et al. *Ann Clin Biochem* 1994; 31:447-54
3. De Blasio P et al, *Prenat Diagn* 1999; 19;499-504

Screening	Markers	Cost (Rs.)	TAT (days)
First trimester	Serum free $\beta$ -hCG	500	4
	Serum free $\beta$ -hCG PAPP-A	1395	6
	Serum free $\beta$ -hCG PAPP-A Fetal Nuchal Translucency (NT)	1395 + charge for NT detection by Ultra sound	6
Second trimester	Serum AFP	500	4
	Serum AFP free $\beta$ -hCG	1000	4
	Serum AFP free $\beta$ -hCG UE3 (TST)	1500	4