DUAL TEST (MATERNAL SERUM SCREEN - 2)

> "Double test is a maternal blood screening test performed in the first trimester between 9-13 weeks of gestation. The specific substances analyzed are: Free Beta hCG, PAPP-A"

Double test is a maternal blood screening test performed in the first trimester between 9-13 weeks of gestation. The specific substances analyzed are: Free Beta hCG, PAPP-A

FreeBeta HCG

Human chorionic gonadotropin (hCG) is a protein made by the placenta that consists of two subunits, alpha and beta. hCG is present in two forms: intact hCG (consisting of both alpha and beta subunits) and free Beta hCG (the beta subunit of hCG alone). Serum levels of hCG tend to be high in patients carrying a fetus affected with Down syndrome. The median concentration for free Beta hCG in the blood of women carrying fetuses with Down syndrome, measured as a multiple of the median (MoM), is 1.9 (compared to 1.0 for unaffected fetuses). This large difference provides the basis for the discrimination between affected and unaffected fetuses. Intact hCG, on the other hand, does not discriminate as well: its median MoM is less than 1.3. Furthermore, free Beta is significantly reduced in the blood of women carrying Trisomy 18 fetuses, with a median MoM of 0.18. Free Beta is the most specific and sensitive marker for Down syndrome and Trisomy 18.

The main advantage of measuring free Beta hCG, instead of intact hCG, is a significantly higher detection rate for a given false positive rate.

PAPP-A

Pregnancy Associated Plasma Protein A (PAPP-A) is also produced by the placenta. PAPP-A is significantly reduced in the blood of women carrying fetuses with Down syndrome and Trisomy 18: the median MoMs are 0.44 and 0.32, respectively (compared to 1.0 for unaffected fetuses). These dramatic differences from normal provide excellent discrimination between affected and unaffected fetuses.

Nuchal Translucency

One of the single most important breakthroughs in chromosome abnormality screening was the ultrasonographic observation of a correlation between a nuchal translucency (accumulation of fluid at the back of the fetal neck) observed on ultrasound between 10 and 13 weeks and the incidence of Down syndrome and other abnormalities. NT is increased in fetuses with chromosome abnormalities, heart defects, and certain genetic syndromes.

Combination of Double test with nuchal translucency increases the detection rate of both Trisomy 21 and 18. This test is performed between 11-13 weeks of gestation.

Dr. Ashish Gupta,
D.Q.M. & Dy. H.O.D.(Hematology & Immunology)
Dr. Lal Path Labs

New Test Introductions at LPL

TEST NAME	SAMPLE TYPE	TAT	MRP
ALDOLASE	2-3ml SERUM	SAME DAY	500
SLA (ANTI SOLUBLE LIVER ANTIGEN)	2-3ml SERUM	SAMPLE BY 7am, REPORT SAME DAY	1000
QUANTIFERON-TB GOLD; GAMMA INTERFERON	1ml WHOLE BLOOD IN SPECIAL TUBE AVAILABLE FROM LPL	SAMPLE BY TUESDAY/ FRIDAY 7am, REPORT ON THURSDAY/ MONDAY	2500
CMV AVIDITY IGG	2-3ml SERUM	SAMPLE BY 3 PM, REPORT SAME DAY	400
TOXOPLASMA AVIDITY IGG	2-3ml SERUM	SAMPLE BY 3 PM, REPORT SAME DAY	400
RUBELLA AVIDITY IGG	2-3ml SERUM	SAMPLE BY 3 PM, REPORT SAME DAY	400
TORCH AVIDITY IGG	2-3ml SERUM	SAMPLE BY 3 PM, REPORT SAME DAY	1000
DUAL TEST (MATERNAL SERUM SCREEN – 2)	2-3ml SERUM	SAMPLE BY 3 PM, REPORT NEXT DAY	1600

QUANTIFERON-TB GOLD ; GAMMA INTERFERON

Usage: Latent tuberculosis infection is a non-communicable asymptomatic condition which might develop into overt Tuberculosis within months or years. QuantiFERON-TB Gold test detects the response to peptide antigens that simulate Mycobacterial proteins through a cell mediated immune reaction. These proteins are absent from BCG strains and most non-tuberculosis Mycobacteria. Individuals infected with M.tuberculosis complex organisms usually have lymphocytes in their blood that recognize these antigens. This recognition process involves the generation and secretion of the cytokine IFN-Gamma. The detection and quantification of IFN-Gamma forms the basis of this test.

Specimen: Collect 1 mL whole blood each in a set of special QuantiFERON – TB Gold IT tubes available from LPL. Shake the tubes vigorously at least 10 times to ensure thorough mixing. Ship at room temperature to reach lab within 16 hours OR Incubate the tubes immediately at 370C for 16-24 hours. Ship refrigerated within 72 hrs. DO NOT FREEZE.

Method: Enzyme Immunoassay

FROM THE EDITOR'S DESK

In this Mega-marathon of life, we sacrifice many vital elements of our being including health. Lifestyle disorders accumulate and small changes manifest as serious ones.

Lifestyle disorders have been the neglected stepchild of primary health care and yet contribute to high levels of medical morbidity and clinically significant decrements in function. In this issue we highlight two such recent reports on lifestyle diseases, deficiency of the "Sunshine hormone Vitamin – D", surprisingly prevalent in a tropical country like ours and Heart disease, high blood pressure and atherosclerosis - conditions that are usually associated with the senior population - creeping into young adulthood.

According to recent pharmaceutical research studies prescription drug use by younger adults for heart disease- related conditions, lipid lowering drugs and antihypertensives, is increasing at a rapid rate, far outpacing older adults and offering a glimpse into the forthcoming clinical and financial challenges facing our health care system. The flip side of the story is that younger patients are taking medications to control conditions that, if left untreated, could lead to heart attacks and strokes - indicating that physicians are screening patients more regularly and treating these precursors more aggressively than in the past. Predictors of future risk of cardiovascular disease can help formulate appropriate strategies for screening and treating young men at heightened risk.

Another offshoot of changing lifestyles is increasing maternal age at first pregnancy and consequently increased risk of fetal chromosomal abnormalities. Considering that an estimated 75 percent of affected fetuses are born to mothers younger than 35 years, it is pertinent to provide pregnant women, younger than 35 years, with noninvasive screening methods. In his article, Dr A. Gupta, details maternal screening methods and first trimester screening – Double Test, recently added to LPL test menu.

It is indeed encouraging to have received a tremendous response to the previous issue of INSIGHT and we continue to look forward to your valuable feedback and suggestions to guide us in addressing the various issues in need of diagnostic service.

Dr. REENA NAKRA,

Chief Editor,

Chief of Lab and Zonal Lab Operations Head –South, East and West Zone

LPL, Noida

E-mail: reena.nakra@lalpathlabs.com

Dr Lal PathLabs
INSIGHT
Volume 10 May. 2008 News Letter

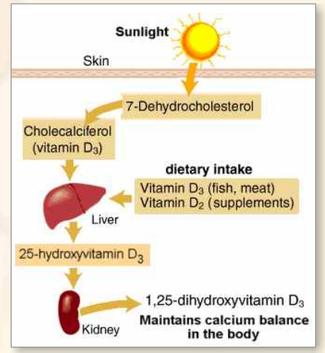
Vitamin D and B12 Deficiency on the Rise in Corporate Executives

Lifestyle disorders are now also manifesting as deficiency of Vitamin B12 and Vitamin D, the sunshine hormone. A recent study, published in the Indian Journal Of Occupational and Environmental Medicine (IOJEM), estimates the deficiency level of Vitamin B12 at nearly 68% and Vitamin D at about 28% among the executives surveyed [1]

The IJOEM study assessed the incidence of vitamin B12 and D3 deficiency among company executives by subjecting them to analysis of blood levels of vitamin D (25 Hydroxy Cholecalciferol) and vitamin B12.

The study targeted corporate executives who

- had vague complaints of unexplained pain in upper and lower limbs
- worked long hours in air-conditioned offices
- and in spite of living in a city with a tropical climate, were barely exposed to sunlight.



Prevalence rate of Vitamin D deficiency and relation to exercise and exposure to sunlight

The IJOEM study concludes that in spite of living in tropical climates with abundant sunlight, vitamin D3 deficiency is evident in the urban Indian population as a corporate lifestyle disorder. Office executives especially, are not exposed to sunlight due to changing lifestyles, long working hours, modern environment and lack of physical exercise or indoor exercises thus hardly exposing themselves to sunlight.

Prevalence rate of B12 deficiency and relation to diet

High incidence of vitamin B12 deficiency was observed probably due to predominently vegetarian diet and insufficient consumption of dairy products, poultry, meat and alcohol consumption. Alcohol is known to retard the absorption of vitamin B12.

Does vitamin D offer benefits beyond bone health?

Researchers continue to report on a growing body of evidence that indicates vitamin D plays a much larger role in regulating health than simply helping build strong bones. Vitamin D appears to help regulate cell growth, immunity, musculoskeletal health and various other biological functions that play a role in keeping us healthy (2-5).

A study in the current issue of the Archives of Internal Medicine reports that men with blood levels below 15 nanograms per milliliter had 2 1/2 times the risk of having a heart attack or dying. Follow up period was 10 years in the study and after additional adjustment for family history of myocardial infarction, body mass index, alcohol consumption, physical activity, history of diabetes mellitus and hypertension, low- and high-density lipoprotein cholesterol levels and triglyceride levels, this relationship remained significant.

Another study found that low levels of vitamin D increased the risk of diabetes, and yet another reasearch linked deficiencies to an increased risk of dying from breast cancer.

Sunlight and diet as sources for Vitamin D:

The skin manufactures the majority of the body's vitamin D by tissue synthesis from cholesterol catalyzed by skin exposure to UV light. But as adults age the ability to make vitamin D through the skin diminishes. Avoiding sun exposure or using sunscreen can also limit a person's production of vitamin D. Adequate sun exposure is 10-20 minutes for people with pale skin in the summer noonday sun. For others with darker skin tones, the corresponding time is 60-100 minutes. Since the length of time for sun exposure varies with geographical location, skin pigmentation, percent body fat and age, careful attention to obtaining dietary sources of vitamin D and daily supplementation is prudent.

Some researchers have reported that vitamin D intake is often too low to sustain healthy circulating levels of the active form of this nutrient. This is mostly due to unique dietary patterns, such as low milk consumption, vegetarian diet, limited use of dietary supplements or low fish intake.

LPL Tests for assessment of Vitamin D and B12 levels

1. 25 hydroxy vitamin D enzyme immunoassay – Vitamin D derived from diet and tissue synthesis is hydroxylated to 25 hydroxy vitamin D. This is the major circulating form of vitamin D. Due to its long half life, the blood levels can be used to assess the vitamin D status of the patients. The levels vary with exposure to sunlight, peaking in summers in ambulatory patients.
2. 1,25 dihydroxy vitamin D enzyme immunoassay – This is the bioactive form of the vitamin formed by hydroxylation in the kidney. The process in the kidney is stimulated by parathyroid hormone and hypophosphatemia. These levels are useful in monitoring patients with renal osteodystrophy, chronic renal failure, parathyroid disease and vitamin D dependence or resistance.
3. Vitamin B12 Cyanocobalamin chemiluminescent immunoassay (CLIA) – Deficiency leads to megaloblastic anemia, neurological changes including demyelination

Sampling Cautions

1. Overnight fasting **2.** Sample should be wrapped in aluminium foil to protect from light **3.** Avoid vitamin supplements 24 hours prior to sampling

References:

1. Incidence of vitamin B12 / D3 deficiency among company executives, Gulvady, Chaitanya; Pingle, Shyam; Shanbhag, Shrinivas; Indian Journal of Occupational and Environmental Medicine, Vol. 11, No. 2, May-August, 2007, pp. 83-85 **2.** Raiten DJ, Picciano MF. Vitamin D and health in the 21st century: bone and beyond. Executive summary. Am J Clin Nutr. 2004;80:1673S-1677S. **3.** Cantorna MT, Zhu Y, Froicu M, Wittke A Vitamin D status, 1,25-dihydroxyvitamin D3, and the immune system. Am J Clin Nutr. 2004;80:1717S-1720S. **4.** Montero-Odasso M, Duque G. Vitamin D in the aging musculoskeletal system: an authentic strength preserving hormone. Mol Aspects Med. 2005;26:203-319. **5.** Holick MF. Vitamin D: importance in the prevention of cancers, type 1 diabetes, heart disease, and osteoporosis. Am J Clin Nutr. 2004;79:362-371.

PREDICTING CARDIOVASCULAR DISEASE IN THE YOUNG

Coronary heart disease remains the leading cause of death and it is emerging more so in the young population.

The South Asian countries of India, Pakistan, Bangladesh, Sri Lanka and Nepal account for about a quarter of the world population and contribute the highest proportion of cardiovascular diseases compared with any other region globally. Deaths related to cardiovascular disease occur 5 to 10 years earlier in South Asian Countries than in Western countries according to a recent study (1).

The researchers found that the prevalence of protective risk factors (leisure time physical activity, regular alcohol intake, and daily intake of fruits and vegetables) were markedly lower in South Asian study participants compared with those from other countries. Also some harmful factors were more common in native South Asians than in individuals from other countries: history of diabetes, current and former smoking, history of hypertension, psychosocial factors such as depression and stress at work or home, and elevated ApoB/ApoA-I ratio (a protein/lipid). This suggests that lifestyle changes implemented early in life have the potential to substantially reduce the risk of AMI in South Asians.

RISK FACTORS

Major coronary disease risk factors, many of which are modifiable, are strong contributors to prediction of future risk, even in young men. These may help in formulating appropriate strategies to identify young men at heightened risk for death from coronary heart disease in later adulthood.

High cholesterol and high blood pressure are two of the leading risk factors for heart disease, heart attack and stroke. High LDL cholesterol can cause atherosclerosis causing narrowing and hardening of the arteries. Hypertension, can weaken the arterial walls and make them more prone to atherosclerosis. Both conditions can lead to thrombi that can block blood flow and result in a heart attack or stroke. Most cases of atherosclerotic vascular disease become clinically apparent in patients aged 40-70 years. Autopsy studies show that coronary atherosclerosis begins as early as 20 years of age and in a recent study found severely stenotic coronary arteries (narrowing more than 40%) in 19% of men in their early thirties.

1. **SERUM CHOLESTEROL** - A recent study demonstrated the ability of serum cholesterol level and other well-known risk factors for coronary heart disease (particularly age, systolic blood pressure, cigarette smoking, and educational level) to predict death from coronary heart disease over 20 years in men 18 to 39 years of age (2). Relative risks for most of the major risk factors were of similar magnitude in young men and middle-aged men, and the relative risk associated with an elevated serum cholesterol level was found to be significantly higher in young men than in middle-aged men. This adds to the evidence that early development of coronary atherosclerosis is associated with risk factors which are largely modifiable. Furthermore, it indicates that it would be worthwhile to assess young people (e.g. for cholesterol) who are genetically predisposed to heart disease so they can make early lifestyle changes if necessary.

   The National Cholesterol Education Program (NCEP) recommends cholesterol screening in all adults 20 years of age or older.

2. **LDL/ Oxidised LDL** - The mechanisms of atherogenesis remain uncertain. The "response to injury" theory is most widely accepted. Probable causes of endothelial injury include LDL cholesterol; infectious agents; toxins, including the by-products of cigarette smoking; hyperglycemia; and hyperhomocystinemia.

   Elevated serum levels of LDL cholesterol overwhelm the antioxidant properties of the healthy endothelium and result in abnormal endothelium metabolism of this lipid moiety.

   Oxidized LDL is capable of a wide range of toxic effects and cell/vessel wall dysfunctions that are characteristically and consistently associated with the development of atherosclerosis. Furthermore, oxidized LDL activates inflammatory processes at the level of gene transcription by up-regulation of nuclear factor kappa-B, expression of adhesion molecules, and recruitment of monocytes / macrophages. Circulating monocytes infiltrate the intima of the vessel wall , and these tissue macrophages act as scavenger cells, taking up LDL cholesterol and forming the characteristic foam cells of early atherosclerosis.

3. **Lipoprotein (a)** - Numerous studies have linked elevated plasma levels of lipoprotein (a), an LDL like moiety that circulates in the blood attached to apolipoprotein (a), with the development of coronary artery disease. This complex shares structural domains with the fibrinolytic enzyme plasminogen and may render the molecule prothrombotic. The LDL like moiety is susceptible to oxidation and may be particularly atherogenic.

4. **High triglycerides** are associated with low (HDL) high density lipoprotein and are a probable risk factor for vascular disease. Recent studies have shown that plasma triglyceride level is a risk factor for cardiovascular disease independent of high-density lipoprotein cholesterol level.

5. **hs-CRP:-** A growing number of studies have examined inflammatory markers as predictors of recurrent CVD and death in different settings, including the short-term risk, long-term risk, and risk after revascularization procedures such as percutaneous coronary intervention (PCI), including the risk of restenosis. Although several markers have been studied, the strongest association with prognosis has been with fibrinogen and hs-CRP.

   hs-CRP consistently predicts new coronary events in patients with unstable angina and acute myocardial infarction.

   For patients with acute coronary syndromes, cutpoints for elevated hs-CRP different than those for prediction in asymptomatic patients may be useful. For example, a level of >10 mg/L in acute coronary syndromes may have better predictive qualities, whereas a level of >3 mg/L may be more useful in patients with stable coronary disease.

   In acute coronary syndromes, hs-CRP predicts recurrent myocardial infarction independent of troponins, which suggests it is not merely a marker for the extent of myocardial damage. Elevated hs-CRP levels also seem to predict prognosis and recurrent events in patients with stroke and peripheral arterial disease. This suggests that hs-CRP may have a role in risk stratification of patients with established CVD.

   Measurement of markers should be done twice (averaging results), optimally two weeks apart, fasting or nonfasting in metabolically stable patients. If hs-CRP level is >10 mg/L, test should be repeated and patient examined for sources of infection or inflammation.

Relative Risk Category and Average hs-CRP Level

Low <1 mg/L

Average 1.0 to 3.0 mg/L

High >3.0 mg/ L

6. **Fibrinogen** - Fibrinogen may be elevated in association with risk factors for atherosclerosis, including smoking, age, and diet. However this is a strong independent predictor of future cardiovascular events in apparently healthy patients.

7. **C-reactive protein** - C-reactive protein levels add to the predictive value of lipid parameters in determining the first myocardial infarction in apparently healthy men and women without a history of coronary heart disease.

   A recent study found CRP concentrations to be higher in healthy Indian Asians than in European whites and was accounted for by greater central obesity and insulin resistance in Indian Asians. The results of the study suggest that inflammation or other mechanisms underlying elevated CRP values may contribute to the increased CHD risk among Indian Asians (3)

8. **Homocysteinemia** - Homozygous hyperhomocysteinemia is associated with extensive atherosclerosis at an early age. Atherogenesis due to hyperhomocysteinemia likely is due to oxidative damage to the endothelium followed by platelet activation and thrombus formation.

9. **Insulin** - Many people with diabetes develop heart disease, but a new study says that determining who has high levels of insulin in the blood -- a condition that precedes diabetes -- may better predict who is at risk for having a heart attack. Measurement of blood glucose and hemoglobin A1c is appropriate in patients with diabetes mellitus (4).

Tests available at Dr. Lal Path Labs:-

Lipid profile basic (cholesterol, Triglyceride ,Direct HDL, LDL ,VLDL)

Lipid profile complete (Cholesterol, Triglyceride, HDL,VLDL,LDL by Electrophoresis, Chol/HDL Ratio,Chylomicrons)

Lipid Profile Comprehensive (Apolipoprotein A1, B & RATIO, Lp (a), Cholesterol, Triglyceride, LDL/HDL ratio,Chol/HDL ratio, Chylomicrons, LDL Subfractions, Homocysteine, Uric Acid, Fibrinogen, Cardio CRP, Plasminogen Activator inhibitor-1 (PAI-1)

Lipid Extended 1 (Apolipoprotein A1, B & ratio,LP(A), Cholesterol, Triglyceride, LDL/HDL ratio,Chol/HDL ratio, Chylomicrons electrophoretically)

Lipid Extended 11 (Apolipoprotein A1, B & ratio, Cholesterol,Triglyceride, LDL/HDL ratio,Chol/HDL ratio,Chylomicrons Electrophretically)

Lipid Profile with LDL Subfraction.

Homocysteine

Cardio CRP

Insulin

Dr. A.K. Kapoor,
Consultant & HOD Front Office, Main Lab

References:

1. JAMA and Archives Journals (2007, January 18). South Asians Have Higher Levels Of Heart Attack Risk Factors At Younger Ages. **2.** EL Navas-Nacher et al. Risk factors for coronary heart disease in men 18 to 39 years of age. Annals of Internal Medicine 2001 134: 433-439. **3.** John C. Chambers et al. C-Reactive Protein, Insulin Resistance, Central Obesity, and Coronary Heart Disease Risk in Indian Asians From the United Kingdom Compared With European Whites ,Circulation. 2001;104:145-150 **4.** American Heart Association (1998, August 14). High Blood Levels Of Insulin Possible Independent Predictor Of Heart Attack Risk.

PRENATAL SCREENING WITH MATERNAL SERUM SCREEN 3 (TRIPLE TEST) AND DOUBLE TEST

Prenatal screening is an issue that has become more important over the past few years. Most elements of standard prenatal care are relatively straightforward and easy for patients to understand and accept, but screening and diagnostic testing for chromosomal abnormalities remain confusing, emotionally charged and fraught with uncertain risks.

The most commonly used test for genetic diagnosis is amniocentesis, but the rate of spontaneous fetal loss related to amniocentesis averages about one in every 200 procedures.

Because of this risk, serum analyte testing has become an important, noninvasive first step in detecting patients at risk for congenital abnormalities. Current maternal serum analyte screening helps identify women at risk for neural tube defects (NTDs), trisomy 21 and trisomy 18.

NTDs are one of the most common serious fetal malformations. The incidence of NTDs is 1-5 for every 1,000 births. These defects include anencephaly, spina bifida and encephalocele. Spina bifida has the third highest lifetime cost of any congenital anomaly.

Trisomy 21 (Down syndrome) is associated with mental retardation, malformation of the heart, gastrointestinal tract, eyes and ears, and early Alzheimer's disease.1 The overall risk of having an affected fetus is one in 1,000 live births.

The second trimester risk is one in 270 in women 35 to 40 years of age, and one in 100 in women older than 40 years.1 It has long been accepted that women who are 35 years or older at the time of delivery should be offered prenatal diagnosis with amniocentesis or chorionic villus sampling.

Although the risk for trisomy 21 increases with maternal age, an estimated 75 percent of affected fetuses are born to mothers younger than 35 years.

Because of this risk, it is important to provide pregnant women who are younger than 35 years with noninvasive screening for this trisomy.

Trisomy 18 (Edwards' syndrome) occurs in one in every 6,000 births and is associated with low birth weight, mental retardation and cranial, cardiac and renal malformations.

Most infants affected with this trisomy die within the first year of life.

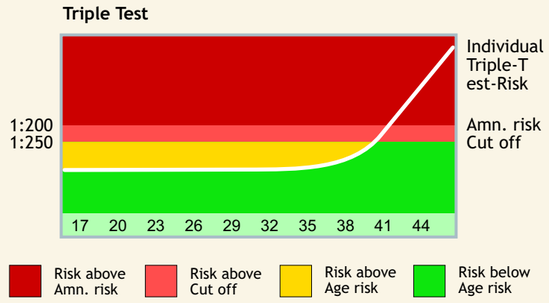
TRIPLE TEST

"Triple test is a maternal blood screening test that looks for three specific substances: AFP, hCG, and Estriol."

ALPHA-FETOPROTEIN: AFP is synthesized in the yolk sac, gastrointestinal tract and liver of the fetus. Fetal plasma levels peak at 10 to 13 weeks' gestation and decline progressively until term, while maternal levels peak in the third trimester. Laboratory measurements of AFP levels are reported as multiples of the median (MOM).

HUMAN CHORIONIC GONADOTROPIN: A complex glycoprotein, hCG is produced exclusively by the syncytiotrophoblast shortly after implantation into the uterine wall. It increases rapidly in the first eight weeks of gestation.9 It then decreases steadily until 20 weeks, when it plateaus. Maternal weight and parity affect hCG levels. An increased hCG level appears to be the most sensitive marker for detecting trisomy 21. A low hCG level is associated with trisomy 18. The hCG levels are normal in NTDs.

UNCONJUGATED ESTRIOL: Unconjugated estriol is produced by the placenta from precursors provided by the fetal adrenal glands and the liver.6 It increases steadily throughout pregnancy to a higher level than is normally produced by the ovaries. Unconjugated estriol levels are decreased in trisomy 21 and trisomy 18.



The triple screen is most accurate if done between 16 and 18 weeks of gestation, but it can be done from 14 to 22 weeks of gestation.

Interpreting Triple Analyte Screening Results

Anomaly	AFP	hCG	uE3
NTDs	Increased	Normal	Normal
Trisomy 21	Decreased	Increased	Decreased
Trisomy 18	Decreased	Decreased	Decreased

Factors affecting the results of Triple tests:

The most important factor affecting the analysis is Maternal age.

Other factors are:

- Maternal weight,
- insulin dependent diabetes mellitus,
- gestation (single, twin or more),
- I.V.F. status. If a pregnancy is through I.V.F., then D.O.B. is taken of the lady whose eggs are used for the procedure.(own or donor).
- race and
- Other factors like smoking also affect the result to a lesser extent.

Reasons to Perform Antenatal Screening

- In most cases, the news will be good and may reassure the patient.
- Some patients may decide to terminate the pregnancy when faced with a lethal abnormality.
- Anomaly detection may allow specialized antenatal treatment and change perinatal treatment.
- If the patient chooses not to terminate the pregnancy, she might still find it reasonable to avoid a cesarean delivery for fetal distress in a child with a lethal anomaly.
- It is much gentler to the parents to learn of anomalies early rather than duringthe stressful, usually happy time of labor and delivery.
- The parents have time to prepare emotionally and financially.
- The family can educate themselves about the anomaly.

Counseling

Beyond the technical aspects of maternal serum analyte screening lies the human aspect. Patients need to understand what screening tests are being offered and how they may affect them. The physician needs to provide patients with the risks and benefits of performing these screens. While appropriate counseling before testing is essential, it is not always done. Counseling should be nondirective and include all relevant information. It is important that patients understand that this is a screening test and that a positive or negative result is not an absolute indication that something is or is not wrong with their infant. It is also important

that patients not be required to make definitive decisions about how they will respond to the results before the testing occurs.

What Additional Tests Will Be Offered?

A woman who receives a positive result should have her due date confirmed by ultrasound, if that was not done before the test was drawn. Incorrect dates may change a positive result to a negative result.

A change in pregnancy dates may also show that the test was drawn too early and will have to be repeated. When pregnancy dates are confirmed, a targeted ultrasound is recommended to look for birth defects and other signs of pregnancy complications.

All cases of anencephaly should be found on ultrasound in the second-trimester. Most cases of spina bifida, but not all, can be seen on ultrasound as well. Ultrasound can also find changes that suggest an increased risk for Down syndrome or trisomy 18; however, it is important to remember that ultrasound cannot diagnose or rule out a chromosome abnormality.

If a chromosome abnormality is suspected, amniocentesis is offered for prenatal diagnosis. Amniocentesis involves the removal of a small amount of amniotic fluid. This fluid naturally contains fetal skin cells that can be studied to find out if a baby has extra or missing chromosomes. The amniotic fluid also contains AFP and a chemical called acetylcholinesterase (AChE) that can be measured to detect neural tube defects.

Dr Lal PathLabs