NAVIGENE

Genetics : Screening • Diagnosis • Research

## Navigene Baby Screen (NBS)

Navigene Baby Screen is a simple, non-invasive and painless test performed to detect 110 serious medical conditions. 48 hrs after a baby is born, through a Urine sample

| Disorders | Symptons & Signs | NBS 110 | NBS 61 | NBS 31 |
|---|---|---|---|---|
| **Amino Acidopathies & Organic Acidemia** | | | | |
| 1 Propionic acidemia (PPA) | Lethargy, poor feeding, hypotonia, Acidosis | Y | Y | Y |
| 2 Multiple carboxylase deficiency | Decreased muscle tone, developmental delay | Y | Y | Y |
| 3 Methylmalonic acidemia (MMA) - Cbl C, D | Poor growth, Micocephaly | Y | Y | Y |
| 4 Methylmalonyl- CoA mutase deficiency (MUT) | Developmental Delay | Y | Y | Y |
| 5 MMA, due to abnormal metabolism, absorption and transport of Vit B12 | Vomitting,Seizures, stroke,encephalopathy | Y | | |
| 6 Malonic Aciduria | Vomitting, diarrhea, seizues | Y | Y | |
| 7 Isobutyryl-CoA dehydrogenase deficiency (IBG) | Poor feeding,failure to thrive, a weakened and enlarged heart. | Y | | |
| 8 2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG) | Loss of motor skills, mental retardation and epilepsy | Y | Y | |
| 9 Methylmalonic semialdehyde dehydrogenase deficiency | Progressive encephalopathy, Convulsions, lethargy | Y | | |
| 10 Beta- ketothiolase deficiency (BKT) | Vomiting, dehydration, trouble breathing, extreme tiredness | Y | Y | Y |
| 11 Isovaleric acidemia | Sweaty feet' odour, poor feeding, seizures | Y | Y | Y |
| 12 3-methylcrotonyl CoA carboxylase deficiency | Acute metabolic acidosis, delayed development | Y | Y | Y |
| 13 3-methylglutaconic aciduria | Cardiomyopathy & skeletal myopathy, delayed growth. | Y | Y | Y |
| 14 Barth Syndrome ( type 2 : 3-methylglutaconic aciduria) | Hypotonia, fatigue, poor apetite,delayed growth | Y | | |
| 15 3-hydroxy-3-methylglutaric aciduria | Severe metabolic acidosis , Developmental delay | Y | Y | Y |
| 16 Glutaric aciduria type II | Sweaty feet' odour, low blood sugar, Enlarged liver and birth | Y | Y | Y |
| 17 Glutaric aciduria type I | Large head, Spasm, jerking, rigidity mnuscle weakness | Y | Y | Y |
| 18 Mevalonic acidemia | Abnormal head shape, delayed development | Y | | |
| 19 Glyceroluria (X-linked disorder) | Vomitting, hypoglycemia, mental retardation | Y | | |
| 20 Phenylketonuria (PKU) | Developmental delay and behaviour problems | Y | Y | Y |
| 21 Benign hyperphenylalaninemia (H-PHE) | Brain Damage | Y | Y | |
| 22 2-Methyl 3-hydroxy butyric aciduria (2M3HBA) | Hypotonia | Y | Y | |
| 23 Tyrosinemia type I | Failure to thrive, Hepatomegaly, Jaundice, Ascites | Y | Y | Y |
| 24 Tyrosinemia type II | Eyes sensitivity to light, Redness in eye, Painful skin lesions on | Y | | |
| | plam and soles, Mental retardation | Y | | |
| 25 Tyrosinemia type III | Mild mental retardation, Convulsions | Y | | |
| 26 Transient tyrosinemia in infancy | Prolonged jaundice, lethargy | | | |
| 27 Tyrosinemia caused by a liver disfunction | Hepatic dysfunction | Y | | |
| 28 Maple syrup urine disease (MSUD) | Sweet smelling, Urine, Poor feeding, seizures, coma | Y | Y | Y |
| 29 N Acetylglutamate synthase Deficiency | Lethargy, Vomitting and Coma | | | |
| 30 Carbamylphosphate synthetase 1- deficiency ** | Lethargy with respiratory problems | Y | Y | |
| 31 Ornithine transcarbamylase (OCT) deficiency | Irritability, seizures, hypotonia, respiratory distress, coma | Y | Y | Y |
| 32 Citrullinemia | Lethargy, poor feeding, vomitting, seizures, loss of consciousness | Y | Y | |
| 33 Citrullinemia type II (CIT II) | Aggression, Hyperactivity, self injuring | Y | Y | |
| 34 Argininosuccinic aciduria | Poorly controlled body Temperature, Coma, Mental retardation, | Y | Y | |
| 35 Argininemia | Loss of developmental milestones, Spasticity, Ataxia, MR | Y | Y | |
| 36 Hypermethioninemia | Neurological problems, motor developmental delay | Y | Y | |
| 37 Homocystinuria | Mental retardation | Y | Y | Y |
| 38 Alkaptonuria | Damage to cartilage | Y | Y | Y |
| 39 Tryptophanuria with dwarfism | Short stature, mental retardation | Y | | |
| 40 Xanthurenic aciduria | Mental retardation | Y | Y | |
| 41 Valinemia | Failure to thrive, Developmental delay | Y | | |
| 42 Hyperleucine- isoleucinemia | Seizures, failure to thrive | Y | | |
| 43 Dihydrolipoyl dehydrogenase(E3) deficiency | Burnt sugar smell to urine and body, M.R. | Y | Y | |
| 44 3- hydroxyisobutyryl-CoA-deacylase deficiency | Hypoketotic hypoglycemia, Neurological damage | Y | | |
| 45 Histidinuria | Mental retardation/ different facial features | Y | Y | Y |
| 46 Hartnup disease | Sensitivity to light and eye defect | Y | | |
| 47 Lysinuric Protein intolerance | Poor weight gain | Y | | |
| 48 Cystinuria | Encephalopathy, deafness, blindness, kidney stones. | Y | | |
| 49 Iminoglycinuria | Mental retardation and kidney problems | Y | Y | |
| 50 2- ketoadipic aciduria | Developmental delay and other neurological problems | Y | Y | Y |
| 51 Saccharopinuria | Dysmorphic features, Short stature | Y | | |
| 52 Hydroxylysinuria | Mental retardation, behavioural problems and hyperactivity | Y | Y | |
| 53 Cystathinionuria ** | Mental retardation | Y | | |
| 54 Hyperprolinemia type I | Neurological or psychiatric problems | Y | Y | |
| 55 Hyperprolinemia type-II ** | Seizures, mental retardation | Y | | |
| 56 Hyper hydroxyprolinemia | Mental retardation | Y | | |
| 57 L-2 hyroxy glutaric aciduria | Hypotonia, vision problem, developmental delay, seizures | Y | | |
| 58 Hawkinsinuria | Small head, lethargy, irritability, failure to thrive | Y | | |
| 59 Biotinidase deficiency | Lack of coordination, learning disabilities, seizure ,muscle | Y | Y | Y |
| 60 Fumarate hydratase deficiency | Seizures with severe retardation | Y | Y | |
| 61 Hyperornithinemia-hyperammoninemia hyperhomocitrullinemia (HHH) | Developmental delay, learning disabilities | Y | Y | Y |
| **Disorders of Sugar Metabolism** | | | | |
| 62 Galactosemia | Jaundice, speech difficulties, learning disabilities, neurological Impairment. | Y | Y | Y |
| 63 Galactokinase deficiency (GALK) | Damage in Liver, Brain, Kidneys, Eyes | Y | | |
| 64 Galactose epimerase deficiency (GALE) | Dysmorphic features | Y | | |
| 65 Transient galactosemia | Dysmorphic features | Y | | |
| 66 Hereditary Fructose Intolerance | Growth abnormalities | Y | | |
| 67 D-glyceric aciduria | Poor weight gain | Y | | |
| 68 Fructose-1,6-diphosphatase deficiency | Hypoglycemia with ketosis | Y | Y | |
| 69 Endogeneous sucrosuria | Mental retardation | Y | | |
| 70 Lactose Intolerance | Slow growth, Weight Loss | Y | Y | |
| **Disorders of Fatty Acid Metabolism** | | | | |
| 71 Short-chain acyl- CoA dehydro- genase deficiency (SCAD) | Low blood sugar, lethargy | Y | Y | |
| 72 Medium-chain acyl- CoA dehydrogenase deficiency (MCAD) | Low blood sugar, Sudden Infant death, Developmental delay | Y | Y | Y |
| 73 Long-chain acyl- CoA dehydrogenase deficiency (LCHAD) | Muscle weakness / Consistent muscle pain | Y | Y | Y |
| 74 2 ketoglutaric aciduria | Hypotonia, Hepatomegaly | Y | Y | Y |
| 75 Ethylmalonic aciduria | Vomitting, lethargy, acidosis, hypoglycemia | Y | | |
| 76 Dicarboxylic Aciduria | Vomitting, poor feeding, muscle weakness, Metabolic acidosis | Y | | |

| Disorders | Symptons & Signs | NBS 110 | NBS 61 | NBS 31 |
|---|---|---|---|---|
| **Peroxisomal Diseases** | | | | |
| 77 Zellweger syndrome | Decreased muscle tone, dysmorphic features | Y | Y | |
| 78 Neonatal adrenoleukodystrophy | Hyperactivity, disruptive behaviour | Y | | |
| 79 Infantile refsum disease (IRD) | Blindness and hearing problems / retinitis pigmentosa | Y | Y | |
| 80 Zellweger like syndrome (ZLS) | Decreased muscle tone, severe psycho-motor retardation | Y | | |
| 81 Primary hyperoxoluria | Kidney failure | Y | Y | |
| **Disorders of purine, pyrimidine metabolism** | | | | |
| 82 Adenosine deaminase deficiency | Episodes of (otitis) ear and upper respiratory tract infections | Y | | |
| 83 Lesch - Nyhan syndrome | Mental retardation, Self biting habit | Y | Y | |
| 84 Partial deficiency of hypoxanthine-guanine phosphoribosyltransferase | Acute inflammatory arthritis | Y<br>Y | | |
| 85 Adenine phosphoribosyl transferase deficiency | Urinary tract infection | Y | Y | |
| 86 Xanthinuria | Acute renal failure | Y | Y | Y |
| 87 Orotic aciduria | Heart malformation and anaemia | Y | Y | |
| 88 Thymine- uraciluria | Mental retardation | Y | | |
| 89 Dihydropyridinase deficiency | Neonatal convulsions | Y | | |
| 90 Hyperuric acidemia | Joint pain | Y | | |
| 91 Beta- Ureido propionase Deficeincy | Hypotonia, dystonic movements, severe developmental delay | | | |
| **Other IEM** | | | | |
| 92 Hyperglycinuria | Failure to thrive | Y | Y | |
| 93 Hypersarcosinemia | Failure to thrive, developmental delays | Y | Y | |
| 94 Imidazole aminoaciduria | Delayed development | Y | | |
| 95 D-2hydroxy glutaric aciduria | Intellectual disability, developmental delay | Y | | |
| 96 Carnosinase deficiency | Decreased muscle tone, delayed development | Y | | |
| 97 Canavan disease | Severe regression of milestones | Y | Y | Y |
| 98 5-oxoprolinuria | Ataxia, spastic, seizures | Y | Y | |
| 99 Glutathionuria | Hemolytic anaemia | Y | | |
| 100 Succinic semialdehyde dehyrogenase deifiency | Fine-motor skills, Learning disability | Y | Y | Y |
| 101 Hyperpipecolatemia | Severe delayed development | Y | Y | |
| 102 Formiminoglutamic aciduria | Intellectual disability, developmental delay | Y | | |
| 103 3-aminoisobutyric aciduria | Neurological impairment | Y | Y | |
| **Lactic acidemia,Hyperpyruvic acidemia** | | | | |
| 104 Pyruvate dehydrogenase (E1) deficiency ** | Lethargy and respiratory problems | Y | | |
| 105 Pyruvate dehydrogenase phosphatase deficiency | Lactic acidosis with decreased muscle tone | Y | | |
| 106 Pyruvate carboxylase deficiency ** | Respiratory problems | Y | Y | Y |
| 107 Pyruvate decarboxylase deficiency | Psychomotor retardation with vision problem | Y | Y | Y |
| 108 Leigh syndrome ** | General weakness with heart problems | Y | | |
| 109 Cytochrome C oxidase deficiency | Generalized weakness , abnormalities of the heart & kidneys. | Y | | |
| 110 Cytochrome aa3-b deficiency (De toni fanconi debre syndrome) | Excessive thirst & urination, Vomitting, seizures | Y | | |
| ** Enzyme assays required | | Y | | |

## Other Packages from Navigene:

Parents/Doctors can opt for these packages in addition to above mentioned packages Sample requirement : Heal Prick Blood Spot on special filter paper

| Test | Disorders Covered |
|---|---|
| NBS-3 | Congenital Hypothyroidism, Congenital Adrenal Hyperplasia, G6PD Deficiency |
| NBS-4 | NBS-3 + Biotinidase |
| NBS-5 | NBS-4 + Galactosemia |
| NBS-6 | NBS-5 + PKU |
| NBS-7 | NBS-6 + MSUD |
| NBS-8 | NBS-7 + Cystic Fibrosis |

**Note : These packages can be customized as per the Hospitals/Doctors requirements**

## About Navigene Genetic Science

Navigene specializes in Genetics and offers screening & diagnostic tests at both prenatal and newborn stages. Tests offered by Navigene are

**New Born Screening :** Urine & Blood test

**Confirmatory testing :** Urine Organic acid Analysis, Enzyme Assays

**Prenatal Screening :** First & Second Trimester Screening

**Prenatal Diagnosis :** Karyotyping on CVS, Amniotic fluid, POC etc Molecular Genetic Testings

## Disclaimer :

1. NBS tests a sample for a maximum of 110 disorders. There are many more genetic metabolic disorders which may lead to similar manifestations in future.

2.NBS does not rule out all metabolic abnormalities/disorders. It is important to monitor baby's mental and physical growth and take a proactive step, should there be any visible sighs and symptoms of the baby not meeting the growth and social milestones.

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