VGENOMICS™

Newborn Screening (NBS) Panels

1- NBS Panel - vNBS-7

This panel covers a spectrum of conditions including the following but not limited to:

- Congenital Hypothyroidism: Essential for identifying thyroid function issues early on to manage growth and cognitive development.
- Congenital Adrenal Hyperplasia: Screening to prevent adrenal crisis and manage hormone levels.
- G6PD Deficiency: Important for detecting this common enzyme deficiency to prevent hemolytic anaemia.
- Galactosemia: Early identification to avoid complications from galactose intake.
- Phenylketonuria (PKU): Detects inability to metabolize phenylalanine to prevent intellectual disability and other health issues.
- Biotinidase Deficiency: Screening to prevent biotin deficiency and its associated symptoms.
- Cystic Fibrosis: Early detection can significantly improve quality of life through management of lung function and nutrition.

By opting for vNBS-7, healthcare providers can ensure comprehensive care for newborns, covering a range of critical conditions with a single, streamlined test.

2 - NBS Panel – vNBS - 4

- Congenital Hypothyroidism (CH)
- Congenital Adrenal Hyperplasia (CAH)
- G6PD Deficiency
- Phenylketonuria (PKU)

3 - NBS Panel – vNBS – 6

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- G6PD Deficiency
- Phenylketonuria (PKU)
- Galactosemia (GALT)
- Biotinidase Deficiency

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4 - NBS Panel – vNBS - 10+ (11 Disorders Covered)

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- G6PD Deficiency
- Phenylketonuria (PKU)
- Galactosemia
- Biotinidase Deficiency
- Sickle Cell Anemia (Hb SS): Identifying this genetic disorder to initiate early intervention strategies.
- Sickle Cell Disease (Hb S/C): Screening for this variant of sickle cell disease to provide timely management and care.
- Beta Thalassemia: Detecting this blood disorder early to prevent serious complications through proper treatment and monitoring.
- Variant Hemoglobinopathies including Hb E (Var Hb): Covering a range of variant hemoglobin disorders to ensure comprehensive hemoglobinopathy screening.

5 - NBS Panel – vNBS - 55+ (58 Disorders Covered)

Amino Acid Disorders (19 Disorders)

This panel covers a spectrum of conditions including the following but not limited to:

- Argininemia (ARG)
- Citrullinemia Types I & II (CIT-1, CIT-II
- Maple Syrup Urine Disease (MSUD)
- Tyrosinemia Types I-III (TYRI, TYRII, TYR III)
- Homocystinuria (HCY)
- Disorders related to the biopterin cofactor (BIOPT)

Fatty Acid Oxidation Disorders (33 Disorders)

A range of disorders including but not limited to:

- Carnitine Acylcarnitine Translocase Deficiency (CACT)
- Medium and Very Long Chain Acyl-CoA Dehydrogenase Deficiencies (MCAD, VLCAD)
- Propionic Acidemia (PROP)
- Multiple Acyl-CoA Dehydrogenase Deficiency (GA-II)
- Methylmalonic Acidemia variants (Cbl A, B, Cbl C, D)
- Specific enzyme deficiencies (e.g., HMG, 3MCC)

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Others

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- G6PD Deficiency
- Galactosemia
- Biotinidase Deficiency
- Cystic Fibrosis

6 - NBS Panel – vNBS - 120*

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- G6PD deficiency
- Hemoglobinopathies
- Amino Acid Disorders (42 Disorders)
- Carbohydrate Disorders (8 Disorders)
- Fatty Acid Oxidation Disorders (12 Disorders)
- Peroxisomal diseases (4 Disorders)
- Disorders of purine, pyrimidine metabolism (3 Disorders)
- Organic Acid Disorders (22 Disorders)
- Urea Cycle Disorders (9 Disorders)
- Mitochondrial disorders (6 Disorders)
- Non-IEM Conditions (7 Disorders)

7 - NBS Panel – vNBS Haemoglobinopathies

- Sickle Cell Anaemia (Hb SS)
- Sickle Cell Disease (Hb S/C)
- Beta Thalassemia
- Variant Hemoglobinopathies including Hb E (Var Hb)

By opting for vNBS-7, healthcare providers can ensure comprehensive care for newborns, covering a range of critical conditions with a single, streamlined test.

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List of diseases covered in the panel:

vNBS - 4

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- GBPD Deficiency
- Phenylketonuriai (PKU)

vNBS - 6

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- G6PD Deficiency
- Galactosemia
- Phenylketonuria (Pku)
- Biotinidase Deficiency

vNBS - 7

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- G6PD Deficiency
- Galactosemia
- Phenylketonuria (PKU)
- Biotinidase Deficiency
- Cystic Fibrosis

vNBS - 10+

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- G6PD Deficiency
- Galactosemia
- Phenylketonuria (PKU)
- Biotinidase Deficiency
- Sickle Cell Anaemia (Hb SS)
- Sickle Cell Disease (Hb S/C)
- Beta Thalassemia
- Variant Hemoglobinopathies Including Hb E (Var Hb)

vNBS - 55+

- Amino Acid Disorders (19 Disorders)
- Fatty Acid Oxidation Disorders (33 Disorders)
- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- G6PD Deficiency
- Galactosemia
- Biotinidase Deficiency
- Cystic Fibrosisipsum

vNBS - 120

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- G6PD Deficiency
- Hemoglobinopathies (4 Disorders)
- Amino Acid Disorders (42 Disorders)
- Carbohydrate Disorders (8 Disorders)
- Fatty Acid Oxidation Disorders (12 Disorders)
- Peroxisomal Diseases (4 Disorders)
- Disorders Of Purine, Pyrimidine Metabolism (3 Disorders)
- Organic Acid Disorders (22 Disorders)
- Urea Cycle Disorder (9 Disorders)
- Mitochondrial Disorders (6 Disorders)
- Non-IEM Conditions (7 Disorders)

vNBS - Haemoglobinopathies

- Sickle Cell Anaemia (Hb SS)
- Sickle Cell Disease (Hb S/C)
- Beta Thalassemia
- Variant Hemoglobinopathies including Hb

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Detailed list of Disorders Covered

vNBS 55+

FATTY ACID OXIDATION DISORDERS

Medium-chain acyl- CoA
Long-chain 3- hydroxyacyl-CoA
Medium-chain ketoacyl- CoA thiolase deficiency (MCKAT)
Carnitine Acylcarnitine Translocase Deficiency CACT
3 - Hydroxy Long Chain Acyl - CoA Dehydrogenase Deficiency LCHAD
Medium Chain Acyl - CoA Dehydrogenase Deficiency MCAD
Neonatal Carnitine Palmitoyl Transferase Deficiency Type II CPT - 11
Very Long Chain Acyl - CoA Dehydrogenase Deficiency VLCAD
Carnitine Palmitoyl Transferase Deficiency Type 11 CPT - 1
2,4 - Dienoyl - CoA Reductase Deficiency1 DE - RED
Multiple Acyl - CoA Dehydrogenase Deficiency GA - II
Short - chain Acyl - CoA Dehydrogenase Deficiency SCAD
Trifunctional Protein Deficiency TFP
Short chain Hydroxy Acyl - CoA Dehydrogenase Deficiency SCHAD
3 - Hydroxy - 3 - Methylglutaryl - CoA Lyase Deficiency HMG
Glutaric Acidemia Type I GA - I
Isobutyryl - CoA Dehydrogenase Deficiency IBG
Isovaleric Acidemia IVA
2 - Methylbutyryl - CoA Dehydrogenase Deficiency 2MBG
3 - Methylcrotonyl - CoA Carboxylase Deficiency 3MCC
3 - Methylglutaconyl - CoA Hydratase Deficiency 3MGA
Methymalonyl - CoA Mutase Deficiency MUT
Maternal Vitamin B12 Deficiency
Mitochondrial Acetoacetyl - CoA Thiolase Deficiency BKT
Propionic Acidemia PROP
Multiple CoA Carboxylase Deficiency MCD
Malonic Aciduria MAL
Hyperalimentation
Medium Chain Triglyceride Oil Administration MCT
Treatment with Benzoate , Pyvalic Acid , or Valproic Acid
Liver Disease
Presence of EDTA Coagulants in Blood Specimen
Carnitine Uptake Deficiency CUD
Medium - chain ketoacyl - CoA thiolase deficiency MCAT
Methylmalonic Acidemia (Cobalamin disorders) Cbl A , B
Methylmalonic Acidemia with homocystinuria Cbl C , D

AMINO ACID DISORDERS

Argininemia ARG
Argininosuccinic Aciduria ASA
5 - Oxoprolinurial 5 - OXO
Carbamoyphosphate Synthetase 1 Deficiency (CPS 1)
Citrulinemia CIT - 1
Homocystinuria HCY
Hypermethioninemia MET
Hyperammonemia , Hyperomnithinemia , Homocitrullinuria Syndrome 1 HHH
Hyperomnithinemia with Gyral Atrophy1 HOGA
Maple Syrup Urine Disease MSUD
Classical Hyperphenylalaninemia
Transient Neonatal Tyrosinemia
Tyrosinemia Type I TYRI
Tyrosinemia Type II TYRII
Tyrosinemia Type II TYR III
Ornithine transcarbamylase deficiency
Citrulinemia type II
Biopterin defect in cofactor biosynthesis BIOPT (BS)
Biopterin defect in cofactor regeneration BIOPT (REG)

Congenital Hypothyroidism
Congenital Adrenal Hyperplasia
G6PD Deficiency
Galactosemia
Biotinidase Deficiency
Cystic Fibrosisipsum

vNBS 120

ORGANIC ACID DISORDERS

Propionic acidemia (PPA)
Multiple carboxylase deficiency (MCO)
Methyimalonic acidemia (MMA)- Cbl C, D
Methyimalonyl- CoA mutase deficiency (MUT)
Methyimalonic aciduria, cbiA and cbiB forms (MMA, Cbl, A,B)
Malonic acidemia (MAL)
Isobutyryl-CoA dehydrogenase deficiency (IBG)
2-Methylbutyryk-CoA dehydrogenase deficiency (2MBG)
Methylmalonic semialdehyde dehydrogenase deficiency
Beta- ketothiolase deficiency (BKT)
Isovaleric acidemia (IVA)
3-methylcrotonyl CoA carboylase deficiency (MCC)
3-methylglutaconic aciduria
3-hydroxy-3-methylglutaric aciduria (HMG CoA lyase deficiency)
Glutaric aciduria type I (GA TypeII)
Glutaric aciduria type II (GA TypeI)
Malonyk-CoA-decarboxylase deficiency
2-Hydroxyglutaric aciduria
Mevalonate kinase deficienc
2- ketoadipic aciduria
Dihydrolipoyl dehydrogenase(E3) deficiency
4-hydroxybutyric aciduria

UREA CYCLE DISORDERS

N-acetyl gutamate synthase deficiency
Carbamylphosphate synthetase 1 (CPS1) deficiency
Ornithine transcarbamylase (OCT) deficiency
Citrullinemia Type I (CIT type I)
Citrullinemia Type II (CIT type II)
Argininosuccinic aciduria (ASA)
Amininemia (ARG)
Hyperomithinemia-hyperammoninemia-hyperhomocitrullinemia (HHH) syndrome
NICCD

MITOCHONDRIAL DISORDERS

Pyruvate dehydrogenase (E1) deficiency
Pyruvate dehydrogenase phosphatase deficiency
Pyruvate carboxylase deficiency
Pyruvate decarboxylase deficiency
Leigh syndrome
Fumarate hydratase deficiency

CARBOHYDRATE DISORDERS

Galactosemia
Galactokinase deficiency (GALK)
Galactose epimerase deficiency (GALE
Transient galactosemia
Fructokinase deficiency
D-glyceric aciduria
Fructose-1,6-diphosphatase deficiency
Endogeneous sucrosuria

NON IEM CONDITIONS

Neuroblastoma
Ketotic Hyperglycinuria
Hyperuric acidemia
Parenteral Feeding, Amino acid infusion, formua Feeding and bloodtransfusion
Medium Chain Trigyceride (MCT) Oil Administration
Anti-convulsant drug metabolites
Poor quality sample e.g. fungal contamination)

AMINO ACID DISORDERS

Phenylketonuria (PKU)
Defects of biopterin cofactor biosynthesis (BIOPT BS)
Defects of biopterin cofactor regeneration (BIOPT REG)
GPT cyclohydrolase (GIPCH) deficiency**
Dihydropteridine reductase deficiency **
Benign hyperphenylalaninemia (H-PHE)
Tyrosinemia type I
Tyrosinemia type II
Tyrosinemia type III
Transient tyrosinemia in infancy
Tyrosinemia caused by a liver disfunction
Maple syrup urine disease (MSUD)
Hypermethioninemia
Homocystiouria
Alkaptonuria
Xanthurenic aciduria
Valinemiaa
3- hydroxyisobutyryl-CoA-deacylase deficiency
Histidinuria
Hartnup disease
Lysinuric Protein intolerance
Familial Renal iminoglycinuria
Iminoglycinuria
Hawkinsinuria
Hydroxylysinuria
Hyperprolinemia type I
Hyperprolinemia type-II **
Hyper hydroxyprolinemia
Cystinuria
Nonketotic Hyperglycinuria (NKHG)
Hypersarcosinemia
Imidazole aminoaciduri
Formiminoglutamic aciduria
Serum camosinase deficiency
Glutathione synthase deficiency
3-aminoisobutyric aciduria
Histidinemia
Biotinidase deficiency
Canavan disease
5-oxoprolinuria
Hyperleucine- isoleucinemi
Hyperomithinuria with Gyrate Atrophy

PEROXISOMAL DISORDERS

Primary hyperoxoluria Type 1
Primary hyperoxoluria Type 2
Adenine phosphoribosy transferase deficiency
Xanthinaria

FATTY ACID DISORDERS

Short-chain acyl- CoA dehydrogenase deficiency (SCAD)
Medium-chain acyl- CoA dehydrogenase deficiency (MCAD)
Long:chain acyl- Co dehydrogenase deficiency (LCAD)
Medium/Short-chain 3-hydroxyacyl-Co dehydrogenase deficiency (SCHAD)
Long-chain 3- hydroxyacyl-CoA dehydrogenas deficiency (LCHAD)
Medium-chain ketoacyl- CoA thiolase deficiency (MCKAT)

Congenital Hypothyroidism
Congenital Adrenal Hyperplasia
G6PD deficiencya