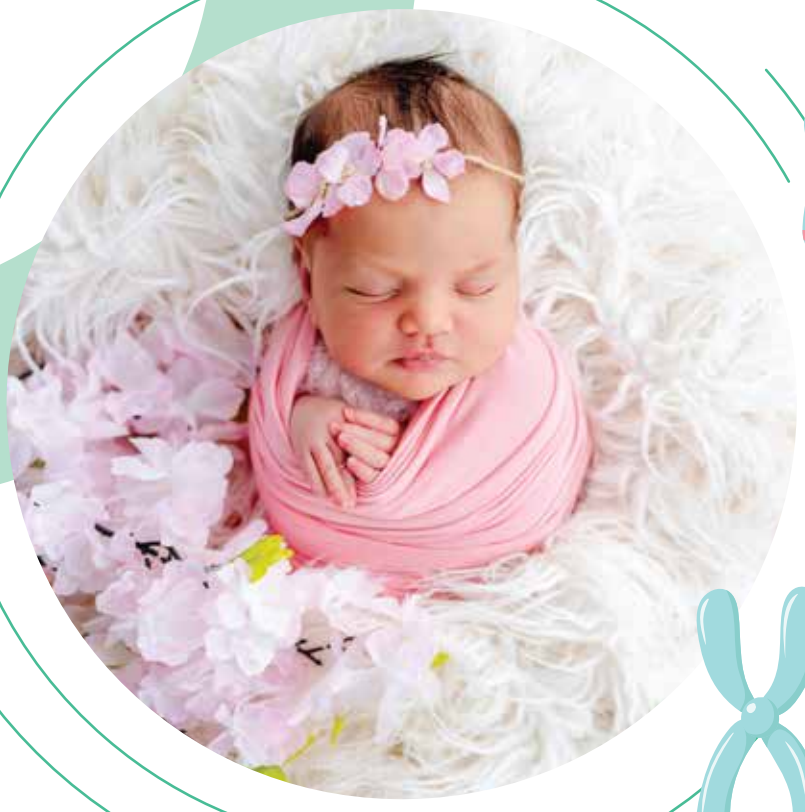

NEW BORN SCREENING

Genetic Analysis Beyond Standard New Born Screening

Rapid Newborn Genetic Screening of

335 Genes by NGS Technology

“New Born Screening” is a Hybridization based library preparation techniques for high-throughput Sequencing

It covers all sequencing of all exonic regions for 335 genes associated with metabolic and genetic diseases. The test is indicated for newborns and children. Offers early screening for genetic and metabolic diseases that appear during the first stages of life, providing key information for disease management and early treatment.

Early detection, intervention & management could prove essential for the infant’s overall health and quality of life.

Potential Benefits

- **Screens for over 300 diseases**
- **Identify potential health risks before symptoms arise**
- **Early detection known to have a positive impact**

- **Expert**
- **Rapid**
- **Accurate**

Panel Performance

Features	Performance
Coverage uniformity	95%
Precision	98%
Reproducibility	98%
Sensitivity	>90%
On Target Ratio	86-93%

Metabolic Disorders

- Amino Acids Metabolic Disorders
- Organic Acid Metabolic Disorders
- Fatty Acid β Oxidation Metabolic Disorders
- Endocrine Disorders
- Carbohydrate Metabolic Disorders
- Metabolic Epilepsy Diseases
- Other Inborn Errors of Metabolism

Genetic Disorders

- Deafness
- Hemophilia B
- B-thalassaemia
- Noonan Syndrome
- Marfan Syndrome

Specimen Required

Dry Blood Spots (size 3.2mm, 5 pieces) by heel prick test

Peripheral Blood (1ml)

genomekundli

This test is designed for

The test is indicated for newborns and children who do not present symptoms of disease.

Neonates with abnormal results of routine biochemical screening, MS/MS screening or failure on routine hearing screenings.

Neonates with clinical manifestation of delayed jaundice, difficulty in feeding, vomiting, diarrhea, anemias.

Advantages

Expert Specially designed gene detection kit for newborn screening

Screen for diseases beyond standard Newborn testing

Accurate Multiple quality control, multi-center verification, Leading phenotypic-genotypic database

Commercial Name	Cat No.
Genome Kundli NGS Panel (New Born Screening)	G2MGK29001-ill G2MGK29001-MG G2MGK29001-TF

S.No.	Disease Name
1	2,4 Dienoyl-CoA reductase deficiency
2	2-methyl-3-hydroxybutyric aciduria
3	2-methylbutyric aciduria
4	2-methylbutyryl-CoA dehydrogenase deficiency
5	3-hydroxy-3-methylglutaric aciduria
6	3-methylcrotonyl-CoA carboxylase 1 deficiency
7	3-methylcrotonyl-CoA carboxylase 2 deficiency
8	3-methylglutaconic aciduria 1
9	3-methylglutaconic aciduria 3
10	3-methylglutaconic aciduria 7
11	3-methylglutaconic aciduria 9
12	3-phosphoglycerate dehydrogenase deficiency
13	6-pyruvoyl-tetrahydropterin synthase deficiency
14	Abetalipoproteinemia
15	Abnormal metabolism thyroid hormone
16	Achalasia-addisonianism-alacrima syndrome
17	Achondroplasia
18	Acrodermatitis enteropathica
19	Acute hepatic porphyria
20	Adrenoleukodystrophy
21	Agammaglobulinemia X-linked
22	Alagille syndrome
23	Alkaptonuria
24	Alpha-mannosidosis
25	Alport syndrome
26	Argininemia
27	Argininosuccinic aciduria
28	Aromatic L-Amino Acid Decarboxylase Deficiency
29	Aspartylglucosaminuria
30	Ataxia with isolated vitamin E deficiency
31	Autosomal dominant and recessive GTP cyclohydrolase I
32	Autosomal dominant hyperinsulinism due to SUR1 deficiency
33	Autosomal recessive 1A deafness
34	Autosomal recessive 1B deafness
35	Autosomal recessive DOPA-responsive dystonia
36	Autosomal recessive Segawa syndrome
37	Autosomal recessive deafness 18a
38	Autosomal recessive deafness 28
39	Autosomal recessive deafness 31
40	Autosomal recessive deafness 79
41	Bare lymphocyte syndrome II
42	Barth syndrome
43	Benign familial infantile seizures
44	Benign familial neonatal epilepsy
45	Beta-ketothiolase deficiency
46	Biotinidase deficiency
47	Carbamoyl-phosphate synthetase 1 deficiency
48	Carnitine palmitoyl transferase 1a deficiency
49	Carnitine palmitoyl transferase 2 deficiency
50	Carnitine-acylcarnitine translocase deficiency
51	Cerebral creatine deficiency 2 syndrome
52	Cerebral creatine deficiency 3 syndrome
53	Cerebral creatine deficiency syndrome 1
54	Cerebral folate transport deficiency
55	Cerebrotendinous xanthomatosis
56	Chronic granulomatous X-linked disease
57	Citrullinemia 1
58	Citrullinemia 2
59	Classic galactosemia
60	Classic homocystinuria
61	Classical congenital adrenal hyperplasia due to 21-hydroxylase deficiency
62	Combined immunodeficiency due to DOCK8 deficiency
63	Combined malonic and methylmalonic acidemia
64	Combined pituitary hormone deficiencies
65	Combined pituitary hormone deficiency
66	Combined pituitary hormone deficiency 2
67	Complementation group B
68	Complementation group C and E
69	Complementation group D
70	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency
71	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency
72	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency
73	Congenital amegakaryocytic thrombocytopaenia
74	Congenital bile acid synthesis defect 1
75	Congenital bile acid synthesis defect 2
76	Congenital bile acid synthesis defect 4
77	Congenital bile acid synthesis defect 6
78	Congenital cyclic neutropenia 1
79	Congenital disorder of glycosylation 1b
80	Congenital hyperinsulinism
81	Congenital hypothyroidism due to thyroid dysgenesis or hypoplasia
82	Congenital hypothyroidism nongoitrous 1
83	Congenital hypothyroidism nongoitrous 4
84	Congenital hypothyroidism nongoitrous 5
85	Congenital hypothyroidism nongoitrous 6
86	Congenital hypothyroidism nongoitrous 7
87	Congenital hypothyroidism nongoitrous 8
88	Congenital hypothyroidism nongoitrous 9
89	Congenital insensitivity to pain with anhidrosis
90	Congenital lipoid adrenal hyperplasia
91	Cori disease
92	Craniometaphyseal dysplasia
93	Crigler-Najjar syndrome 1 and 2
94	Crisponi syndrome
95	Cryopyrin-associated periodic syndrome
96	Cystathioninuria
97	Cystic fibrosis
98	Cystinosis
99	Cystinuria
100	Cytochrome P450 oxidoreductase deficiency
101	Danon disease
102	Deafness 10
103	Deafness 6
104	Deafness 8
105	Developmental and epileptic encephalopathy 6
106	Developmental delay due to ALDH6A1 deficiency
107	Dravet syndrome
108	Duchenne muscular dystrophy
109	Dystonia dopa-responsive due to sepiapterin reductase deficiency
110	Early infantile epileptic encephalopathy
111	Enteropathy
112	Epilepsy vitamin B6-dependent
113	Erytrhopoietic protoporphyria
114	Ethylmalonic encephalopathy
115	Fabry disease
116	Familial Mediterranean fever
117	Familial glucocorticoid deficiency
118	Familial hemophagocytic lymphohistiocytosis
119	Familial hyperaldosteronism I
120	Familial infantile convulsions with paroxysmal choreoathetosis
121	Favism
122	Frasier syndrome
123	Fructose-1,6-bisphosphatase deficiency
124	GLUT1 deficiency syndrome 1
125	GM1-gangliosidosis 1
126	Galactosemia I
127	Galactosemia IV
128	Galactosemia due to galactokinase deficiency
129	Galactosemia due to galactose epimerase deficiency
130	Galactosemia due to mutarotase deficiency
131	Gaucher disease
132	Glutaric Acidemia II
133	Glutathione synthetase deficiency
134	Glycine encephalopathy
135	Glycogen storage Ia disease
136	Glycogen storage disease 0
137	Glycogen storage disease IB and IC
138	Glycogen storage disease II
139	Glycogen storage disease III
140	Glycogen storage disease Ia
141	Glycogen storage disease VI
142	Glycogen storage disease type
143	Growth delay due to insulin-like growth factor I deficiency
144	HMG-CoA synthase 2 deficiency
145	Hearing loss (HL)
146	Hemophilia A
147	Hemophilia B
148	Hereditary amyloidosis transthyretin-related
149	Hereditary angioedema 1 and 2
150	Hereditary fructose intolerance
151	Hereditary hemorrhagic telangiectasia type 1
152	Hereditary hemorrhagic telangiectasia type 2
153	Hermansky-Pudlak syndrome
154	Heterotaxy
155	Holocarboxylase synthetase deficiency
156	Homocystinuria due to methylenete trahydrofolatereductase deficiency
157	Homocystinuria-megaloblastic anemia due to defect in cobalamin metabolism cblG
158	Homocystinuria-megaloblastic anemia due to defect in cobalamin metabolism cblE
159	Hunter disease
160	Hurler-Scheie syndrome
161	Hyperammonemia due to N-acetylglutamate synthase deficiency
162	Hypercholesterolemia
163	Hyperinsulinemic hypoglycemia 4
164	Hyperinsulinemic hypoglycemia 6
165	Hyperinsulinism-hyperammonemia syndrome
166	Hypermethioninemia due to adenosine kinase deficiency
167	Hypermethioninemia due to glycine N-methyltransferase deficiency
168	Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency
169	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
170	Hyperprolinemia 1
171	Hyperprolinemia II
172	Hypervalinemia and hyperleucine-isoleucinemia
173	Hypocalciuric hypercalcemia I
174	Hypophosphatasia
175	Hypophosphatemic rickets with hypercalciuria
176	Immune dysregulation
177	Immunodeficiency 18
178	Immunodeficiency 19
179	Immunodeficiency by defective expression of MHC class I
180	Isobutyryl-CoA dehydrogenase deficiency
181	Isovaleric acidemia
182	Jervell and Lange-Nielsen syndrome
183	Juvenile polyposis syndrome
184	Krabbe disease
185	L-2-hydroxyglutaric aciduria
186	Leigh syndrome
187	Lipoprotein lipase deficiency (LPL)
188	Loeys-Dietz syndrome
189	Long-chain 3-hydroxyacyl-Coa dehydrogenase (subunit A) deficiency
190	Long-chain 3-hydroxyacyl-Coa dehydrogenase (subunit B) deficiency
191	Lysinuric protein intolerance
192	Lysosomal acid lipase deficiency
193	MHC class II deficiency
194	Malonic aciduria
195	Malonyl-CoA decarboxylase deficiency
196	Maple syrup urine Ia disease
197	Maple syrup urine Ib disease
198	Maple syrup urine disease type III
199	Marfan syndrome
200	Maroteaux-Lamy disease
201	Medium chain Acyl-CoA dehydrogenase deficiency
202	Menkes disease

S.No.	Disease Name
203	Metachromatic leukodystrophy
204	Methionine adenosyltransferase deficiency
205	Methylmalonic Acidemia and Homocysteinemia cblX
206	Methylmalonic acidemia and homocysteinemia cblX
207	Methylmalonic acidemia cblA
208	Methylmalonic acidemia cblB
209	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency
210	Methylmalonic acidemia due to methylmalonyl-CoA mutase deficiency
211	Methylmalonic aciduria
212	Methylmalonic aciduria and homocystinuria cb1J
213	Methylmalonic aciduria and homocystinuria cblC
214	Methylmalonic aciduria and homocystinuria cblD
215	Methylmalonic aciduria and homocystinuria cblF
216	Methylmalonic aciduria due to transcobalamin receptor defect
217	Mevalonic aciduria
218	Mild non-BH4-deficient hyperphenylalaninemia
219	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
220	Morquio A syndrome
221	Morquio B syndrome
222	Mucolipidosis II
223	Mucopolysaccharidosis 7
224	Mucopolysaccharidosis I
225	Mucopolysaccharidosis II
226	Mucopolysaccharidosis IIIA
227	Mucopolysaccharidosis IIIC
228	Mucopolysaccharidosis IIID
229	Mucopolysaccharidosis IVA
230	Mucopolysaccharidosis VI
231	Multiple acyl-CoA dehydrogenase deficiency
232	Multiple endocrine neoplasia I
233	Neonatal diabetes mellitus
234	Neonatal hyperparathyroidism
235	Neonatal lactic acidosis with methylmalonic aciduria
236	Nephrogenic diabetes insipidus 1
237	Nephrogenic diabetes insipidus 2
238	Neu-Laxova syndrome-2
239	Neurofibromatosis type 1
240	Neurofibromatosis type 2
241	Neuronal ceroid lipofuscinosis 2
242	Nevoid basal cell carcinoma syndrome
243	Niemann-Pick A and B disease
244	Niemann-Pick C disease
245	Nonketotic hyperglycinemia
246	Nonsyndromic hearing loss
247	Noonan syndrome
248	Ocular albinism type I
249	Oculocutaneous albinism type IV
250	Optic atrophy 1
251	Ornithine aminotransferase deficiency
252	Ornithine transcarbamylase deficiency
253	Osteogenesis imperfecta
254	Osteopetrosis
255	PSPH - phosphoserine phosphatase
256	Paroxysmal exertion-induced dyskinesia
257	Paroxysmal nocturnal hemoglobinuria
258	Pendred syndrome
259	Peutz-Jeghers syndrome
260	Phenylketonuria
261	Pituitary hormone deficiency 1
262	Polycystic kidney and hepatic disease
263	Polycystic kidney disease
264	Polyendocrinopathy
265	Pompe disease
266	Primary hyperoxaluria 1
267	Primary hyperoxaluria 2
268	Primary hyperoxaluria 3
269	Propionic acidemia
270	Pseudohypoaldosteronism 1
271	Pterin-4-alpha-carbinolamine dehydratase deficiency

S.No.	Disease Name
272	Pyridoxamine 5-prime-phosphate oxidase deficiency
273	Pyridoxine-dependent epilepsy
274	Pyruvate carboxylase deficiency
275	Pyruvate kinase deficiency
276	Q-dihydropteridine reductase deficiency
277	RPE65-associated retinal dystrophy
278	Retinitis pigmentosa 39
279	Retinoblastoma
280	Sanfilippo A
281	Sanfilippo B
282	Sanfilippo C
283	Sanfilippo D
284	Scheie syndrome
285	Sensorineural hearing loss
286	Severe Combined Immunodeficiency
287	Severe Combined Immunodeficiency JAK3
288	Severe combined immunodeficiency Athabaskan
289	Severe combined immunodeficiency B RAG-deficient
290	Severe combined immunodeficiency RAG-deficient
291	Severe combined immunodeficiency X-linked
292	Severe combined immunodeficiency due to IL-7Ralpha deficiency
293	Severe combined immunodeficiency due to adenosine deaminase deficiency
294	Severe congenital neutropenia 3
295	Shah-Waardenburg syndrome
296	Short chain Acyl-CoA dehydrogenase deficiency
297	Sickle cell anemia
298	Sitosterolemia
299	Sly syndrome
300	Spherocytosis
301	Spinal muscular atrophy I
302	Spinal muscular atrophy II
303	Spinal muscular atrophy III
304	Spinal muscular atrophy IV
305	Systemic primary carnitine deficiency
306	Tay-Sachs disease
307	Thrombotic thrombocytopenic purpura
308	Thyroid hormonogenesis 1 defect
309	Thyroid hormonogenesis 2A defect
310	Thyroid hormonogenesis 3 defect
311	Thyroid hormonogenesis 4 defect
312	Thyroid hormonogenesis 5 defect
313	Thyroid hormonogenesis 6 defect
314	Transcobalamin II deficiency
315	Transient infantile liver failure
316	Triple A syndrome
317	Tuberous sclerosis complex 1
318	Tuberous sclerosis complex 2
319	Tyrosinemia 1
320	Tyrosinemia 2
321	Tyrosinemia 3
322	Usher syndrome 1b
323	Usher syndrome 1c
324	Usher syndrome 1d
325	Usher syndrome 1g
326	Usher syndrome 2a
327	Usher syndrome 2d
328	Very long chain Acyl-CoA dehydrogenase deficiency
329	Vitamin D hydroxylation-deficient rickets I
330	Vitamin D-dependent rickets 2
331	Von Hippel-Lindau syndrome
332	Waardenburg syndrome
333	Wilson disease
334	Wolman disease
335	X-linked adrenal hypoplasia congenita
336	X-linked central congenital hypothyroidism with testicular enlargement
337	X-linked hyper IgM syndrome
338	X-linked hypophosphatemic rickets
339	Xeroderma pigmentosum
340	β-thalassaemia

Genes2Me Pvt. Ltd.

1105, 11th Floor, Tower B, SAS Tower, Medicity,
Sector - 38, Gurgaon - 122 001, Haryana, India
Tel : + 91 18001 214030 / + 91 88000 23600
E-mail : contact@genes2me.com www.genes2me.com