MINKOFF CENTER FOR JEWISH GENETICS

**Universal Panel | Fundamental Plus Panel | Fundamental Panel**
**175 Conditions**

1. Congenital Adrenal Hyperplasia, CYP21A2-Related
2. Familial Hyperinsulinism, ABCC8-Related
3. Andermann Syndrome
4. Alpha-Mannosidosis
5. Alpha-Sarcoglycanopathy
6. Alpha Thalassemia, HBA1/HBA2-Related
7. Argininosuccinic Aciduria
8. Autosomal Recessive Spastic Ataxia Of Charlevoix-Saguenay
9. Aspartylglucosaminuria
10. Ataxia With Vitamin E Deficiency
11. Ataxia-Telangiectasia
12. Autoimmune Polyglandular Syndrome Type 1
13. Beta-Sarcoglycanopathy
14. Biotinidase Deficiency
15. Bloom Syndrome
16. Canavan Disease
17. Carnitine Palmitoyltransferase IA Deficiency
18. Carnitine Palmitoyltransferase II Deficiency
19. Cartilage-Hair Hypoplasia
20. Cystic Fibrosis
21. CLN3-Related Neuronal Ceroid Lipofuscinosis
22. CLN5-Related Neuronal Ceroid Lipofuscinosis
23. CLN8-Related Neuronal Ceroid Lipofuscinosis
24. Cohen Syndrome
25. Congenital Disorder Of Glycosylation Type Ia
26. Congenital Disorder Of Glycosylation, MPI-Related
27. Nephrotic Syndrome, NPHS1-Related
28. Cystinosis
29. Familial Dysautonomia
30. Familial Mediterranean Fever
31. Fanconi Anemia, FANCC-Related
32. Free Sialic Acid Storage Disorders
33. Gaucher Disease
34. GJB2-Related DFNB1 Nonsyndromic Hearing Loss And Deafness
35. Glutaric Acidemia, GCDH-Related
36. Glycogen Storage Disease Type Ia
37. Glycogen Storage Disease Type Ib
38. Pompe Disease
39. Glycogen Storage Disease Type III
40. GNE Myopathy
41. BCS1L-Related Disorders

42. Hereditary Fructose Intolerance
43. Homocystinuria, CBS-Related
44. Primary Hyperoxaluria Type 1
45. Primary Hyperoxaluria Type 2
46. Isovaleric Acidemia
47. Krabbe Disease
48. Leigh Syndrome, French-Canadian Type
49. FKRP-Related Disorders
50. Dihydrolipoamide Dehydrogenase Deficiency
51. HADHA-Related Disorders
52. Maple Syrup Urine Disease Type Ib
53. Medium Chain Acyl-CoA Dehydrogenase Deficiency
54. Megalencephalic Leukoencephalopathy With Subcortical Cysts
55. Metachromatic Leukodystrophy
56. Mucolipidosis IV
57. Mucopolysaccharidosis Type I
58. Mucopolysaccharidosis Type IIIA
59. POMGNT-Related Disorders
60. Niemann-Pick Disease Type C1
61. Nijmegen Breakage Syndrome
62. Pendred Syndrome
63. D-Bifunctional Protein Deficiency
64. Phenylalanine Hydroxylase Deficiency
65. Autosomal Recessive Polycystic Kidney Disease, PKHD1-Related
66. PPT1-Related Neuronal Ceroid Lipofuscinosis
67. Combined Pituitary Hormone Deficiency, PROP1-Related
68. Pycnodysostosis
69. Rhizomelic Chondrodysplasia Punctata Type 1
70. Short-Chain Acyl-CoA Dehydrogenase Deficiency
71. Sjogren-Larsson Syndrome
72. Smith-Lemli-Opitz Syndrome
73. SLC26A2-Related Disorders
74. TPP1-Related Neuronal Ceroid Lipofuscinosis
75. Tyrosine Hydroxylase Deficiency
76. Tyrosinemia Type I
77. Usher Syndrome Type 3
78. PCDH15-Related Disorders
79. Wilson Disease
80. X-Linked Juvenile Retinoschisis
81. Peroxisome Biogenesis Disorder Type 1
82. Galactosemia
83. Bardet-Biedl Syndrome, BBS1-Related
84. Bardet-Biedl Syndrome, BBS10-Related
85. Junctional Epidermolysis Bullosa, LAMA3-Related
86. Junctional Epidermolysis Bullosa, LAMB3-Related
87. Junctional Epidermolysis Bullosa, LAMC2-Related
88. Hypophosphatasia
89. Spinal Muscular Atrophy
90. Dysferlinopathy

91. NEB-Related Nemaline Myopathy
92. Joubert Syndrome 2
93. Citrullinemia Type 1
94. Primary Carnitine Deficiency
95. Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency
96. Nephrotic Syndrome, NPHS2-Related
97. Costeff Optic Atrophy Syndrome
98. Niemann-Pick Disease, SMPD1-Related
99. FKTN-Related Disorders
100. Hexosaminidase A Deficiency
101. Hb Beta Chain-Related Hemoglobinopathy
102. Congenital Adrenal Hyperplasia, CYP11B1-Related
103. HMG-CoA Lyase Deficiency
104. 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency
105. Adenosine Deaminase Deficiency
106. X-Linked Adrenoleukodystrophy
107. COL4A4-Related Alport Syndrome
108. COL4A3-Related Alport Syndrome
109. X-Linked Alport Syndrome
110. Alstrom Syndrome
111. Argininemia
112. Bardet-Biedl Syndrome, BBS2-Related
113. Bardet-Biedl Syndrome, BBS12-Related
114. Cerebrotendinous Xanthomatosis
115. Lysosomal Acid Lipase Deficiency
116. Lipoid Congenital Adrenal Hyperplasia
117. Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)
118. RTEL1-Related Disorders
119. Fabry Disease
120. Familial Hyperinsulinism, KCNJ11-Related
121. Fanconi Anemia Complementation Group A
122. Glycine Encephalopathy, AMT-Related
123. Glycine Encephalopathy, GLDC-Related
124. GLB1-Related Disorders
125. Holocarboxylase Synthetase Deficiency
126. Mucopolysaccharidosis Type II
127. Hydrolethalus Syndrome
128. Muscular Dystrophy, LAMA2-Related
129. TGM1-Related Autosomal Recessive Congenital Ichthyosis
130. Gamma-Sarcoglycanopathy
131. Calpainopathy
132. Delta-Sarcoglycanopathy
133. Maple Syrup Urine Disease Type II
134. Maple Syrup Urine Disease Type Ia
135. MKS1-Related Disorders
136. Methylmalonic Acidemia, CblB Type
137. Methylmalonic Aciduria And Homocystinuria, CblC Type
138. Methylmalonic Acidemia, CblA Type
139. Methylmalonic Acidemia, MMUT-Related

140. GNPTAB-Related Disorders
141. Mucolipidosis III Gamma
142. Neuronal Ceroid Lipofuscinosis, CLN6-Related
143. Niemann-Pick Disease Type C2
144. ATP7A-Related Disorders
145. Ornithine Transcarbamylase Deficiency
146. Autosomal Recessive Osteopetrosis Type 1
147. Primary Hyperoxaluria Type 3
148. PCCA-Related Propionic Acidemia
149. PCCB-Related Propionic Acidemia
150. Pyruvate Carboxylase Deficiency
151. Sandhoff Disease
152. Mucopolysaccharidosis Type IIIB
153. Mucopolysaccharidosis Type IIIC
154. X-Linked Severe Combined Immunodeficiency
155. Spastic Paraplegia Type 15
156. Spondylothoracic Dysostosis
157. Tyrosinemia Type II
158. USH2A-Related Disorders
159. MYO7A-Related Disorders
160. USH1C-Related Disorders
161. Xeroderma Pigmentosum Group C
162. Xeroderma Pigmentosum Group A
163. Peroxisome Biogenesis Disorder Type 6
164. Peroxisome Biogenesis Disorder Type 4
165. Peroxisome Biogenesis Disorder Type 3
166. Peroxisome Biogenesis Disorder Type 5
167. Carbamoylphosphate Synthetase I Deficiency
168. EVC-Related Ellis-Van Creveld Syndrome
169. EVC2-Related Ellis-Van Creveld Syndrome
170. Galactokinase Deficiency
171. X-Linked Adrenal Hypoplasia Congenita
172. ERCC6-Related Disorders
173. ERCC8-Related Disorders
174. Congenital Disorder Of Glycosylation Type Ic
175. X-Linked Myotubular Myopathy