Carrier Screening Invitae Broad Panel

114 enfermedades analizadas genéticamente.

1. ABCA3-related conditions (ABCA3)
2. ABCC8-related conditions (ABCC8)
3. Achromatopsia (CNGB3)
4. AHI1-related conditions (AHI1)
5. Aicardi-Goutieres syndrome 2 (RNASEH2B)
6. Alpha-N-acetylgalactosaminidase deficiency (NAGA)
7. Alpha-thalassemia (HBA1/HBA2)
8. Argininosuccinate lyase deficiency (ASL)
9. ARX-related conditions (ARX)
10. Aspartylglucosaminuria (AGA)
11. Atransferrinemia (TF)
12. Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia (AIRE)
13. BBS1-related conditions (BBS1)
14. BBS2-related conditions (BBS2)
15. Beta-ketothiolase deficiency (ACAT1)
16. Biotin-responsive basal ganglia disease (SLC19A3)
17. Bloom syndrome (BLM)
18. Canavan disease (ASPA)
19. Cardioencephalomyopathy (SCO2)
20. Carnitine palmitoyltransferase II deficiency (CPT2)
21. CC2D2A-related conditions (CC2D2A)
22. CEP290-related conditions (CEP290)
23. Cerebrotendinous xanthomatosis (CYP27A1)
24. CFTR-related conditions (CFTR)
25. Citrullinemia type 1 (ASS1)
26. CLN3-related conditions (CLN3)
27. CLRN1-related conditions (CLRN1)
28. Cobalamin C deficiency (MMACHC)
29. Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (CYP21A2)
30. Congenital adrenal insufficiency (CYP11A1)
31. Congenital disorder of glycosylation type Ia (PMM2)
32. Congenital hydrocephalus-1 (CCDC88C)
33. Congenital myasthenic syndrome (CHRNE)
34. Congenital nephrotic syndrome type 1 (NPHS1)

35. Congenital nephrotic syndrome type 2 (NPHS2)
36. DHDDS-related conditions (DHDDS)
37. Dihydrolipoamide dehydrogenase deficiency (DLD)
38. DMD-related conditions (DMD)
39. Donnai-Barrow syndrome (LRP2)
40. DYNC2H1-related conditions (DYNC2H1)
41. Dystrophic epidermolysis bullosa (COL7A1)
42. ERCC2-related conditions (ERCC2)
43. EVC2-related conditions (EVC2)
44. Fabry disease (GLA)
45. Factor IX deficiency (hemophilia B) (F9)
46. Familial dysautonomia (ELP1)
47. Familial hemophagocytic lymphohistiocytosis type 2 (PRF1)
48. Fanconi anemia type C (FANCC)
49. FMR1-related conditions including fragile X syndrome (FMR1)
50. Fraser syndrome (GRIP1)
51. Galactosemia (GALT)
52. GBA-related conditions (GBA)
53. GBE1-related conditions (GBE1)
54. GJB2-related conditions (GJB2)
55. Glycogen storage disease type Ia (G6PC)
56. Glycogen storage disease type II (Pompe disease) (GAA)
57. GNPTAB-related conditions (GNPTAB)
58. HBB-related hemoglobinopathies (HBB)
59. Hereditary fructose intolerance (ALDOB)
60. Hermansky-Pudlak syndrome type 1 (HPS1)
61. Hermansky-Pudlak syndrome type 3 (HPS3)
62. Homocystinuria due to cystathionine beta-synthase deficiency (CBS)
63. Hypophosphatasia (ALPL)
64. Joubert syndrome and related disorders (TMEM216)
65. Krabbe disease (GALC)
66. L1 syndrome (L1CAM)
67. Maple syrup urine disease type 1A (BCKDHA)
68. Maple syrup urine disease type 1B (BCKDHB)
69. Medium-chain acyl-CoA dehydrogenase deficiency (ACADM)
70. Megalencephalic leukoencephalopathy with subcortical cysts 1 (MLC1)
71. Metachromatic leukodystrophy (ARSA)

72. Methylmalonic acidemia (MUT)
73. Mucolipidosis type IV (MCOLN1)
74. Mucopolysaccharidosis type I (IDUA)
75. Muscular dystrophy-dystroglycanopathy (FKRP)
76. Muscular dystrophy-dystroglycanopathy (FKTN)
77. MVK-related conditions (MVK)
78. Myotonia congenita (CLCN1)
79. Nemaline myopathy 2 (NEM)
80. Niemann-Pick disease types A and B (SMPD1)
81. NR0B1-related conditions (NR0B1)
82. Oculocutaneous albinism type 2 OCA2)
83. Oculocutaneous albinism types 1A and 1B (TYR)
84. Opitz GBBB syndrome (MID1)
85. Ornithine transcarbamylase deficiency (OTC)
86. PCDH15-related conditions (PCDH15)
87. PEX7-related conditions (PEX7)
88. Phenylalanine hydroxylase deficiency (PAH)
89. PLP1-related conditions (PLP1)
90. POLG-related conditions (POLG)
91. Polycystic kidney disease (PKHD1)
92. Pontocerebellar hypoplasia type 6 (RARS2)
93. Primary hyperoxaluria type 1 (AGXT)
94. Primary microcephaly (MCPH1)
95. SLC26A2-related conditions (SLC26A2)
96. SLC26A4-related conditions (SLC26A4)
97. SLC37A4-related conditions (SLC37A4)
98. Smith-Lemli-Opitz syndrome (DHCR7)
99. Spinal muscular atrophy (SMN1)
100. Spinocerebellar ataxia (ANO10)
101. Tay-Sachs disease (HEXA)
102. Trimethylaminuria (FMO3)
103. Tyrosinemia type I (FAH)
104. USH2A-related conditions (USH2A)
105. Very long-chain acyl-CoA dehydrogenase deficiency (ACADVL)
106. Vitamin D-dependent rickets type 1A (CYP27B1)
107. Wilson disease (ATP7B)
108. X-linked adrenoleukodystrophy (ABCD1)

109. X-linked creatine transporter deficiency (SLC6A8)
110. X-linked juvenile retinoschisis (RS1)
111. X-linked severe combined immunodeficiency (IL2RG)
112. Xeroderma pigmentosum complementation group C (XPC)
113. Zellweger spectrum disorder (PEX1)
114. Zellweger spectrum disorder (PEX6)