

1. Achondroplasia (dwarfism)
2. Albinism
3. Adrenoleukodystrophy ALD (Lorenzo's Oil)
4. Alzheimer's
5. Amyotrophic lateral sclerosis (ALS)/Lou Gehrig's Disease
6. Bloom's Syndrome
7. Breast Cancer
8. Brittle Bone Disease
9. Canavan disease
10. Coffin-Lowry Syndrome
11. Colorblindness
12. Cystic Fibrosis
13. Down Syndrome
14. Duchenne and Becker Muscular Dystrophy
15. Fragile X Syndrome

16. Galactosemia
17. Gaucher Disease
18. Hemophilia
19. Hereditary Deafness
20. Huntington's Disease
21. Klinefelter Syndrome
22. Krabbe Disease
23. Marfan Syndrome (Alton Giant)
24. Neurofibromatosis
25. Phenylketonuria PKU
26. Polycystic Kidney Disease
27. Prader-Willi Syndrome
28. Progeria (premature aging)
29. Rett Syndrome
30. Sickle-Cell Disease

31. Tay Sachs Disease
32. Trisomy 13 (Patau Syndrome)
33. Trisomy 18 (Edward's Syndrome)
34. Xeroderma pigmentosum
35. Triple X syndrome
36. Turner's syndrome
37. Type 1 diabetes
38. Velocardiofacial Syndrome
39. Waardenburg Syndrome
40. Williams Syndrome Chromosomal Translocation