

Appendix VIII

Sex-linked Genetic Diseases⁵²

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| 1. Addison's disease with cerebral sclerosis | 37. Hypophosphataemic rickets |
| 2. Adrenoleucodystrophy | 38. Ichthyosis (steroid sulphatase deficiency) |
| 3. Adrenal hypoplasia | 39. Incontinentia pigmenti (x-linked dominant, male lethal) |
| 4. Agammaglobulinaemia, Bruton type | 40. Kallmann syndrome |
| 5. Agammaglobulinaemia, Swiss type | 41. Keratosis follicularis spinulosa |
| 6. Albinism, ocular | 42. Lesch-Nyhan syndrome (hypoxanthine-guanine-phosphoribosyl transferase deficiency) |
| 7. Albinism-deafness syndrome | 43. Lowe (oculocerebrorenal) syndrome |
| 8. Aldrich syndrome | 44. Macular dystrophy of the retina |
| 9. Alport syndrome | 45. Menkes syndrome |
| 10. Amelogenesis imperfecta, hypomaturation type | 46. Mental retardation, FMRI type |
| 11. Amelogenesis imperfecta, hypoplastic type | 47. Mental retardation, FRAXE type |
| 12. Anaemia, hereditary hypochromic | 48. Mental retardation, MRXI type |
| 13. Angiokeratoma (Fabry's disease) | 49. Microphthalmia with multiple anomalies (Lenz syndrome) |
| 14. Cataract, congenital | 50. Mucopolysaccharidosis II (Hunter syndrome) |
| 15. Cerebellar ataxia | 51. Muscular dystrophy, Becker type |
| 16. Cerebral sclerosis, diffuse | 52. Muscular dystrophy, Duchenne type |
| 17. Charcot-Marie-Tooth peroneal muscular atrophy | 53. Muscular dystrophy, Emery-Dreifuss type |
| 18. Choroideraemia | 54. Myotubular myopathy |
| 19. Choroidoretinal degeneration | 55. Night blindness, congenital stationary |
| 20. Coffin-Lowry syndrome | 56. Norrie's disease (pseudoglioma) |
| 21. Colour blindness, Deutan type | 57. Nystagmus, oculomotor or 'jerky' |
| 22. Colour blindness, Protan type | 58. Ornithine transcarbamylase deficiency (type I hyperammonaemia) |
| 23. Diabetes insipidus, nephrogenic | 59. Orofaciodigital syndrome (type I, x-linked dominant, male lethal) |
| 24. Diabetes insipidus, neurohypophyseal | 60. Perceptive deafness, with ataxia and loss of vision |
| 25. Dyskeratosis congenita | 61. Perceptive deafness, DNFZ type |
| 26. Ectodermal dysplasia, anhidrotic | 62. Phosphoglycerate kinase deficiency |
| 27. Ehlers-Danlos syndrome, type V | 63. Phosphoribosylpyrophosphate (PRPP) synthetase deficiency |
| 28. Faciogenital dysplasia, (Aarskog syndrome) | 64. Reifenstein syndrome |
| 29. Focal dermal hypoplasia (x-linked dominant, male lethal) | 65. Retinitis pigmentosa |
| 30. Glucose 6-phosphate dehydrogenase deficiency | 66. Retinoschisis |
| 31. Glycogen storage disease, type VIII | 67. Spastic paraplegia |
| 32. Gonadal dysgenesis (XY female type) | 68. Spinal muscular atrophy |
| 33. Granulomatous disease (chronic) | 69. Spondyloepiphyseal dysplasia tarda |
| 34. Haemophilia A | 70. Testicular feminization syndrome |
| 35. Haemophilia B | 71. Thrombocytopenia, hereditary |
| 36. Hydrocephalus (aqueduct stenosis) | 72. Thyroxine-binding globulin, absence or variants of |
| | 73. Xg blood group system |
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⁵² Schedule 2 to the Ordinance