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Autore	Aitken Kenneth J
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Nota di contenuto	An A-Z of Genetic Factors in Autism: A Handbook for Parents and Carers; Acknowledgements; Prelude; Introduction; SECTION A: Focus on the autistic spectrum disorders; Why you might need a book like this; Help and treatment: does one size fit all?; ASD and 'inborn errors of metabolism'; Biochemical individuality: are we all the same?; Is ASD getting more common?; A brief history of ASD research; Early presenting features of ASDs; Physical checklist of features that can be seen on physical examination and which may have clinical relevance; Further clinical aspects that may require investigation Genetic clinical conditions linked with autism 'Alternative' approaches; SECTION B: Clinical disorders seen in the autistic spectrum disorders; 1. 15q11-q13 duplication; 2. Chromosome 2q37 deletion; 3. XXY syndrome; 4. XYY syndrome; 5. 10p terminal deletion; 6. 45,X/46,XY mosaicism; 7. 22q13 deletion syndrome; 8. Aarskog syndrome; 9. Adenylosuccinate lyase (ADSL) deficiency; 10. Adrenomyeloneuropathy (AMN); 11. Angelman syndrome (AS); 12. Apert syndrome; 13. ARX gene mutations; 14. Autism secondary to autoimmune lymphoproliferative syndrome (ALPS); 15. Bannayan-Riley-Ruvalcaba syndrome (BRRS) 16. Basal cell naevus syndrome (BCNS)17. Biedl-Bardet syndrome (BBS); 18. CATCH 22; 19. Cortical dysplasia-focal epilepsy (CDFE) syndrome;

20. CHARGE syndrome; 21. Coffin-Lowry syndrome (CLS); 22. Coffin-Siris syndrome (CSS); 23. Cohen syndrome; 24. Cole-Hughes macrocephaly syndrome (CHMS); 25. Congenital adrenal hyperplasia (CAH); 26. Cowden syndrome (CS); 27. De Lange syndrome (CdLS); 28. Juvenile dentatorubral-pallidoluysian atrophy (JDPLA); 29a. DiGeorge syndrome I (phenotypic overlap); 29b. DiGeorge syndrome II (DGS II); 30. Dihydropyrimidine dehydrogenase (DPYS) deficiency
31. Down syndrome (DS)32. Dravet's syndrome; 33. Duchenne (DMD) and Becker (BMD) muscular dystrophy; 34. Ehlers-Danlos syndrome (EDS); 35. Fragile-X syndrome (FRAX); 36. Fragile-X permutation (partial methylation defects); 37. GAMT deficiency (guanidinoacetate methyltransferase deficiency); 38. Goldenhar syndrome; 39. HEADD syndrome; 40. L-2-hydroxyglutaric aciduria (L-2 HGAA); 41. Hyper IgE syndrome with autism (HiES); 42. Hypomelanosis of Ito (HI); 43. Hypothyroidism; 44. Joubert syndrome; 45. Kleine-Levin syndrome; 46. Lujan-Fryns syndrome; 47. 2-methylbutyryl-CoA dehydrogenase deficiency
48. Mobius/Mobius/Moebius syndrome 49. Myhre syndrome; 50. Myotonic dystrophy (MD1); 51. Neurofibromatosis type 1 (NF1); 52. Noonan syndrome (NS); 53. NAPDD; 54. Oculocutaneous albinism (OCA); 55. Ornithine carbamyltransferase deficiency (OCTD); 56. Orstavik 1997 syndrome; 57. Phenylketonuria (PKU); 58. Pituitary deficiency; 59. Port-wine facial staining and autism; 60. Potocki-Lupski syndrome (PTLS); 61. Prader-Willi syndrome (PWS); 62. Proteus syndrome; 63a. Rett syndrome (RTT); 63b. Rett syndrome (Hanefeld variant) (RSHV); 64. Rubinstein-Taybi syndrome; 65. Schindler disease 66. Smith-Lemli-Opitz syndrome (SLOS)

Sommario/riassunto

The book covers the main genetic disorders associated with autism, including those linked to growth differences, neurodevelopmental problems, gastrointestinal disturbances epilepsy and many others. It alphabetically lists the conditions next to information about how common they are, causes, signs, symptoms, and methods of treatment and management.