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| Nota di contenuto | 1. Cerebrotendinous Xantomatosis (CTX): general review and history of the research on its pathogenesis and therapy -- 2. CerebroTendinous Xanthomatosis (CTX) in Italy -- 3. CTX in The Netherlands -- 4. CTX in Brazil -- 5. Cerebrotendinous xanthomatosis in France -- 6.CTX in Japan -- 7. Cerebrotendinous Xanthomatosis: remaining challenges and the need for international collaboration -- 8. Genetics of CTX -- 9. General Overview Biochemistry with Focus on Clinical Laboratory -- 10. Focus on CTX Cases Identified with Atypical Biochemistry -- 11. Cholestanol accumulation in the brain and eye in cerebrotendinous xanthomatosis -- 12. Role of Bile Acid Pathway Intermediates in Pathology of CTX -- 13. Overview of Neuroimaging in Cerebrotendinous Xanthomatosis Diagnosis -- 14. Novel tools for neuroimaging in cerebrotendinous xanthomatosis -- 15. Epidemiology of Cerebrotendinous Xanthomatosis -- 16. Chenodeoxycholic acid |

treatment in the italian cohort of CTX patients -- 17. Long-term treatment effect in CTX depends on age at treatment -- 18.Can we improve outcomes? Lessons taken from the Israeli CTX Cohort -- 19. Treatment during Pregnancy in CTX -- 20. Cerebrotendinous xanthomatosis (CTX) presenting as neonatal cholestasis, its diagnosis and treatment with primary bile acids, cholic and chenodeoxycholic acids -- 21. Cholic acid treatment in adults with cerebrotendinous xanthomatosis -- 22. Gene Therapy for CTX -- 23. CTX Pediatric presentation -- 24. Ophthalmic manifestation in CTX -- 25. Update on Newborn Screening for Cerebrotendinous Xanthomatosis -- 26. Early treatment improves outcomes for patients with cerebrotendinous xanthomatosis (CTX) -- 27. Cerebrotendinous Xanthomatosis Patient and Family Opinions and International CTX Patient Advocacy.

Sommario/riassunto

This book represents an update on the clinical, biochemical and molecular genetic findings of a rare neurometabolic disorder. Cerebrotendinous xanthomatosis (CTX), first described in 1937, is a rare genetic disorder of bile acid synthesis that can cause developmental and cognitive disability, irreversible neurological damage and premature death. Left untreated, the syndrome is slowly progressive but can now be considered a treatable rare neurologic disease of lipid and bile metabolism. Early identification of this disorder, for example with newborn screening, would be highly significant as the burden of disease if left untreated can be profound, and morbidity and mortality easily prevented through early detection and treatment. The book collects the experience of expert physician-scientists and researchers from all over the world, beginning with the history of the disease and providing updates on the new molecular genetic data, the therapies and the international patient advocacy organizations, also with chapters reporting the experiences of the patients and calling for improved international basic research and clinical collaboration, aiming to advance understanding and enhance outcomes for CTX patients and their families, and foster patient group partnerships. The volume stems from the proceedings of an international meeting held in Jerusalem, Israel, where this disease was investigated for many years by a pioneer in the field, Prof. Vladimir Berginer at the Ben-Gurion University of the Negev. This meeting was hosted by the CTX Alliance, a patient advocacy group based in the USA.