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| 1. Record Nr.           | UNINA9910688389103321  |
| Titolo                  | Clinical and Basic Aspects of Neurofibromatosis Type 1 / / edited by Juichiro Nakayama, Yuichi Yoshida   |
| Pubbl/distr/stampa      | London : , : IntechOpen, , 2022<br>©2022   |
| Descrizione fisica      | 1 online resource (144 pages)  |
| Disciplina              | 616.8  |
| Soggetti                | Clinical neuropsychology   |
| Lingua di pubblicazione | Inglese  |
| Formato                 | Materiale a stampa   |
| Livello bibliografico   | Monografia   |
| Nota di bibliografia    | Includes bibliographical references and index.   |
| Sommario/riassunto      | Neurofibromatosis type 1 (NF1), also known as von Recklinghausen disease, is a major monogenic neurocutaneous disorder. The NF1 gene encodes the protein neurofibromin whose dysfunction promotes tumorigenesis in central and peripheral neuronal tissues. In addition to inducing the formation of cutaneous pigmented lesions or neurofibromas, NF1 affects multiple organ systems, resulting in neurological and psychiatric disorders, orthopedic conditions, and impaired endocrine functions. This book examines the fundamental, clinical, and basic aspects of NF1 over three sections and nine chapters. Topics addressed include bone lesions in children with NF1, diffuse neurofibromatous tissue, seizures in adults with NF1, Ras-GAP function of neurofibromin, endocrine disorders characteristic of NF1, and more. |