Record Nr. UNINA9910461504003321 Autore Featherstone Katie Titolo Creating conditions: the making and remaking of a genetic syndrome / / Katie Featherstone and Paul Atkinson Abingdon, Oxon;; New York, N.Y.:,: Routledge,, 2012 Pubbl/distr/stampa **ISBN** 1-283-45811-X 9786613458117 1-134-00480-X 0-203-87815-9 Descrizione fisica 1 online resource (141 p.) Collana Genetics and society Altri autori (Persone) AtkinsonPaul <1947-> Disciplina 616.85/884 Soggetti Rett syndrome Genetic disorders Electronic books. Lingua di pubblicazione Inglese **Formato** Materiale a stampa Livello bibliografico Monografia Description based upon print version of record. Note generali Includes bibliographical references (p. [123]-126) and index. Nota di bibliografia Includes bibliographical references and index. Nota di contenuto Front Cover; Creating Conditions; Copyright Page; Contents; Acknowledgments; 1. Multiple sites of a syndrome; 2. Making medical entities; 3. The culture of the clinic; 4. The transformation of patienthood; 5. Transforming Rett syndrome; 6. The making and remaking of medical classifications; References; Index Sommario/riassunto "Based on original ethnographic research with scientists, clinicians and families, this book examines Rett syndrome to illuminate more general issues concerning the construction and interpretation of diseases and syndromes. It derives from research with a specialist team of clinicians and scientists, and a series of families referred with a potential diagnosis of Rett syndrome, and documents the scientific, clinical, patient and family experiences over a three-year period. Although Rett syndrome itself is rare, it is one of some 2,000 such syndromes, and its genetic basis has recently been linked to the much broader Autism spectrum. From a sociological or anthropological point of view, it is also of considerable interest as a clinical entity that is undergoing

transformation in the light of recent post-genomic research.

Traditionally, such syndromes have been diagnosed clinically, but increasingly genetic technologies are having an impact on the diagnosis, description and classification of conditions. Rett Syndrome is thus a key exemplar of the implications of genetic medicine that are far-reaching and extend well beyond this particular syndrome"--