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The Journal of Mental Science
Encyclopedia of Ethical, Legal, and Policy Issues in Biotechnology
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connecting an abnormal physical exam to a possible genetic condition is a daunting and inexact task for any physician be they a primary care provider non geneticist specialist or fellowship trained geneticist comprising 31 clinical protocols from the world s foremost clinical geneticists signs and symptoms of genetic conditions provides a practical manual for the diagnosis and management of common human genetic conditions based on their presenting signs and or symptoms each chapter examines a specific clinical finding and leads the user through a step by step approach to a differential diagnosis to maximize clinical utility this handbook features prominent flow chart diagrams that graphically depict the diagnostic approach concise recommendations for laboratory and or imaging studies health supervision and management strategies for the most common conditions associated with each presenting sign or symptom whether for the student resident or seasoned clinician signs and symptoms of genetic conditions will serve as a frontline resource for navigating differential diagnosis

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this volume explains the biology and genetics of asd and provides clinicians and researchers with a comprehensive summary of each genetic factor including the research that links it to asd diagnosis and treatment issues and related animal models as well as detailing relevant professional organisations and avenues for further research

a history of genetic testing warns that such tests may tell us more than we want to know medical geneticists began mapping the chromosomal infrastructure piece by piece in the 1970s by focusing on what was known about individual genetic disorders five decades later their infrastructure had become an edifice for prevention allowing today's expecting parents to choose to test prenatally for hundreds of disease specific mutations using powerful genetic testing platforms in life histories of genetic disease andrew j hogan explores how various diseases were made genetic after 1960 with the long term aim of treating and curing them using gene therapy in the process he explains these disorders were located in the human genome and became targets for prenatal prevention while the ongoing promise of gene therapy remained on the distant horizon in narrating the history of research that contributed to diagnostic genetic medicine hogan describes the expanding scope of prenatal diagnosis and prevention he draws on case studies of prader willi fragile x digeorge and velo cardio facial syndromes to illustrate that almost all testing in medical genetics is inseparable from the larger and increasingly big data oriented aims of biomedical research hogan also reveals how contemporary genetic testing infrastructure reflects an intense collaboration among cytogeneticists molecular biologists and doctors specializing in human malformation hogan critiques the modern ideology of genetic prevention which suggests that all pregnancies are at risk for genetic disease and should be subject to extensive genomic screening he examines the dilemmas and ethics of the use of prenatal diagnostic information in an era when medical geneticists and biotechnology companies have begun offering whole genome prenatal screening essentially searching for any disease causing mutation hogan's focus and analysis is animated by ongoing scientific and scholarly debates about the extent to which the preventive focus in contemporary medical genetics resembles the aims of earlier eugenicists written for historians sociologists and anthropologists of science and medicine as well as bioethics scholars physicians geneticists and families affected by genetic conditions life histories of genetic disease is a profound exploration of the scientific culture surrounding malformation and mutation

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preimplantation diagnosis of genetic diseases a new technique in assisted reproduction editors yury verlinsky and anver m kuliev for individuals with hereditary genetic disorders such as cystic fibrosis muscular dystrophy or hemophilia the decision to reproduce can be a profoundly difficult and emotional problem for specialists in reproductive medicine this situation can raise a myriad of complex often conflicting ethical questions recently a revolutionary approach to screening for genetic disorders has emerged offering unique possibilities for resolving this dilemma preimplantation diagnosis of genetic diseases a new technique in assisted reproduction details the latest procedures for detecting congenital disease in gametes prior to fertilization or in embryos fertilized in vitro prior to uteral implantation this book covers current methods used in oocyte and preembryo sampling as well as the latest techniques in dna and cytogenetic analysis in preimplantation development this text presents detailed practical information on such crucial topics as blastomere and blastocyst biopsy for preimplanation genetic analysis oocyte retrieval and embryo transfer for in vitro fertilization preparation of necessary media and micromanipulation tools polymerase chain reaction techniques for dna analysis of polar bodies and preembryos preimplantation cytogenetic and enzymatic analyses ethical issues involved in preimplantation genetic screening serving as both a practical manual for current methods and a reference for future research and clinical applications preimplantation diagnosis of genetic diseases a new technique in assisted reproduction provides a complete overview of this dramatic development in reproductive medicine this book will be of interest to specialists in fertility medicine obstetrics and gynecology embryology genetics molecular biology and cytogenetics whose work defines this new frontier in biomedical science

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