The Fragile X Syndrome Sourcebook

A Comprehensive Guide for Parents, Caregivers and Families



The Official Parents Sourcebook On Fragile X Syndrome A Directory For The Internet Age

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created for parents who have decided to make education and research an integral part of the treatment process Although it also gives information useful to doctors caregivers and other health professionals it tells parents where and how to look for information covering virtually all topics related to lesch nyhan syndrome also Hereditary Hyperuricemia and Choreoathetosis Syndrome HG PRT deficiency syndrome HGPRT Absence of HPRT Absence of Hyperuricemia Choreoathetosis Self multilation Syndrome Hyperuricemia Oligophrenia from the essentials to the most advanced areas of research The title of this book includes the word official This reflects the fact that the sourcebook draws from public academic government and peer reviewed research Selected readings from various agencies are reproduced to give you some of the latest official information available to date on lesch nyhan syndrome Given parents increasing sophistication in using the Internet abundant references to reliable Internet based resources are provided throughout this sourcebook Where possible guidance is provided on how to obtain free of charge primary research results as well as more detailed information via the Internet E book and electronic versions of this sourcebook are fully interactive with each of the Internet sites mentioned clicking on a hyperlink automatically opens your browser to the site indicated Hard copy users of this sourcebook can type cited Web addresses directly into their browsers to obtain access to the corresponding sites In addition to extensive references accessible via the Internet chapters include glossaries of technical or uncommon terms Children with Fragile X Syndrome Jayne Dixon Weber, 2000 Fragile X syndrome is believed to be the most common genetic cause of mental retardation even more common than Down syndrome It can result in a wide range of developmental delays learning disabilities and physical characteristics which all tend to be more pronounced in boys than in girls At last there is a comprehensive book on fragile X syndrome for parents The first and only book of its kind this book provides a complete sensitive introduction to fragile X syndrome an inherited genetic condition caused by a mutation on the X chromosome This new guide is written by an experienced caring group of parents doctors therapists and teachers who offer an in depth look at the issues and concerns affecting children and Understanding Fragile X Syndrome Isabel Fernández Carvajal, David Aldridge, 2011-08-15 Fragile X their families syndrome is one of the main causes of child developmental delay and autism spectrum disorders This book breaks down the complex science of this genetic disorder and provides the facts and advice that every bewildered parent or professional needs to support individuals with Fragile X syndrome Fragile X Fred Julia Bassell, Jayne Weber, Jeannie Visootsak, 2015-08-10 Look around you Every person you meet has been molded by an intertwining of genetic information and personal experience Fragile X syndrome is a genetic disorder that is not widely understood This book aims to help people of all ages understand that although individuals with fragile X syndrome do have certain challenges these challenges do not define them This book is a valuable tool for educators siblings parents and children with fragile X syndrome *The Source for Fragile X Syndrome* Fragile X Syndrome Handbook Vincent Jerry, 2023-10-26 Fragile X syndrome Gail Harris-Schmidt, Dale Eugene Fast, 2004 FXS is a genetic disorder that results in a variety of intellectual and developmental impairments It is one of the most

prevalent inherited causes of intellectual impairment and autism spectrum disorder Here are a few important details about Fragile X syndrome FXS is caused by a mutation in the FMR1 gene which is located on the X chromosome The mutation occurs on one of the two X chromosomes that are typically present in humans The mutation causes a deficiency in FMRP Fragile X Mental Retardation Protein a protein that is essential for brain development and function Intellectual and Developmental Disabilities Fragile X syndrome is frequently accompanied by a spectrum of intellectual and developmental disabilities These can range in severity but frequently include learning difficulties cognitive impairment and speech and language delays Behavioral and Emotional Difficulties FXS is also associated with behavioral and emotional difficulties Individuals with Fragile X syndrome may exhibit social anxiety repetitive behaviors hyperactivity impulsivity and sensory processing difficulties Some people may also suffer from mood disorders such as melancholy and anxiety Although there are no distinct physical characteristics associated with FXS some individuals may have subtle facial characteristics or connective tissue problems In general these physical characteristics are less pronounced than those seen in other genetic syndromes Autism Spectrum Disorder ASD Many FXS patients also meet the diagnostic criteria for autism spectrum disorder ASD This means they may exhibit impaired social interactions communication difficulties and repetitive behaviors comparable to those observed in autism The severity of FXS can differ significantly between affected individuals Others may have intellectual and developmental disabilities of a more severe nature The diagnosis of Fragile X syndrome is typically confirmed by genetic testing which searches for mutations in the FMR1 gene Also available is prenatal testing for families with a history of the disorder Treatment and Support Although there is no cure for FXS early intervention and a multidisciplinary approach to treatment can assist affected individuals in reaching their maximum potential Individuals with FXS frequently receive speech therapy occupational therapy and behavioral interventions to address their unique requirements To manage symptoms such as anxiety and hyperactivity medications may also be prescribed Families of individuals with Fragile X syndrome may benefit from gaining access to support services and resources such as special education programs support groups and organizations devoted to FXS research and advocacy It is essential to note that FXS affects individuals differently and each individual with the syndrome may experience unique challenges and strengths Early diagnosis and appropriate interventions can make a substantial difference in the lives of individuals and families affected by Fragile X syndrome *The Fragile X Child* Bettv B. Schopmeyer, Fonda Lowe, 1992 Designed primarily for speech language pathologists and occupational therapists this book will also be helpful for teachers parents and educational advocates in identifying children with the fragile X syndrome and in approaching intervention

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