

## Review Article

# Birth defects: Risk factors and consequences

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**Abstract.** Birth defects (BDs) or congenital anomalies include all structural and functional alterations in embryonic or fetal development resulting from genetic, environmental or unknown causes, which result in physical and/or mental impairment. BDs occur in about 3% of newborn babies and in most cases of pregnancy loss. BDs are a very complex and heterogeneous group of single or multiple changes that, in most cases, are of unknown etiology. Among the risk factors are advanced maternal and paternal ages, parental consanguinity, teratogenic agents such as infectious agents and drugs, and poor nutrition, in particular folic acid deficiency. One of the consequences of these defects is the high death rate within the first year of life. Information on BDs is becoming increasingly more important throughout the world so that preventive measures can be taken. Knowledge of BDs enables the development of therapeutic and preventive strategies besides adequate genetic counseling.

**Keywords:** Congenital anomalies, congenital malformation, chromosomal abnormalities

## 1. Classification and etiology

Congenital anomalies or birth defects (BDs) include all functional and structural alterations in embryonic or fetal development resulting from genetic, environmental or unknown causes that originate during pregnancy, are present at birth and that cause physical or mental impairment [1].

BDs are a very complex and heterogeneous group of single or multiple changes that, in most cases, are of unknown etiology. The genetic etiology of BDs is relatively well known and includes monogenic causes (about 20% of cases), chromosomal alterations (approximately 25%), which may be numerical or structural and multifactorial causes (about 50% of cases). The etiology of environmental factors responsible for

disruptive effects exists in approximately 5% of cases and these cases are the most difficult to investigate. Aneuploidy is the cause of BDs in more than 30% of all cases; most are monosomy of chromosome X and trisomies of chromosomes 16, 21 and 22, and are responsible for about 60% of gestational losses. Although trisomies of chromosomes 13, 18 and 21 are responsible for many pregnancy losses, they also have an impact at birth, as they are present in approximately 0.3% of all births together with trisomy of chromosome X [2–4].

Thus, genetic factors are among the most common causes of BDs and are one of the main causes of infant mortality. BDs are observed in about 3% of newborns and are the cause of 50% of spontaneous neonatal deaths [5–8].

Stillbirths occur in between 1 in 1000 and 1 in 160 pregnancies and are related to maternal, fetal, and placental causes; in half of the cases, no cause can be determined. BDs occur in 1–4% of these cases with genetic diseases being a common cause [9,10]. Stillbirths can

\*Corresponding author: Camila Ivo Ferreira Oliveira, Av. Brigadeiro Faria Lima, 3544, São José do Rio Preto – SP, Brazil CEP: 15.090-000. Tel.: +55 17 210 3000; Fax: +55 17 3233 6534; E-mail: camilivo.oliveira@hotmail.com

# **Birth Defects Risks And Consequences**

**United States. Congress. House.  
Committee on Science and  
Technology. Subcommittee on the  
Environment and the Atmosphere**

## **Birth Defects Risks And Consequences:**

Birth Defects Sally Kelly,1976      **Mortality and Maldevelopment** Harold Kalter,2009-02-28 Congenital malformations are worldwide occurrences striking in every condition of society These severe physical abnormalities which are present at birth and affecting every part of the body happen more often than usually realized once in every 33 births The most common after heart defects are those of the neural tube the brain and spinal cord which happen in as many as one in every 350 births They have been noted as curiosities in man and beast throughout recorded history and received great attention in our time by various fields of study for example their faulty prenatal development by embryologists familial patterns by geneticists causation by environmentalists and variability by population scientists Attention turned much in recent years to the relation of these malformations to deficiency of a particular dietary ingredient folic acid a subject this book analyzes in depth The greatest conundrum of all which this latest matter like so much else hinges on is the amazing fact of the tremendous almost universal decrease in the frequency of these anomalies since early in the 20th century The puzzle is What can this downward trend possibly mean and at bottom Whether it is part of a long term cyclical pattern This fascinating biological phenomenon is explored in the book together with various other topics      **Selected References on Environmental Quality as it**

**Relates to Health** ,1976 Monthly Bibliography of MEDLARS based journal articles that describe perturbations in the ecosystems important to health For the most part genetic and clinical literature not included Index medicus format author subject sections      *Consanguinity - Its Impact, Consequences and Management* Lutfi A. Jaber ,Gabrielle J. Halpern,2014-05-23 Consanguineous marriages have been practiced for hundreds of years predominantly by Moslems Although it is generally accepted among communities that the social advantages outweigh other the rate of congenital malformations and genetic diseases among the offspring borne of consanguineous marriages is higher and an increase in sterility rates of abortions stillbirths and neonatal deaths has been suggested by some researchers These problems result in a substantial economic burden within the communities involved in the practice Strategies for reducing the frequency of consanguineous marriages include expansion of educational programs promotion of genetic testing services and research to identify the genes responsible for genetic defects Nowadays with the migration of large numbers of Moslems towards Western countries consanguinity is becoming a practical issue in places where it was barely known previously This eBook serves as a guide for health care workers and counselors of the phenomenon and its associated problems This eBook begins with an introduction of the topic from a social legal and biological perspective It then progresses to research about associated genetic disorders reproductive and social awareness issues through the proceeding chapters The eBook concludes with 2 chapters exploring strategies to counter the effects of consanguinity within the general population      *Centers for Disease Control Birth Defects Study* United States. Congress. House. Committee on Veterans' Affairs. Subcommittee on Hospitals and Health Care,1984      *Long-Term Health Consequences of Exposure to Burn Pits in Iraq and Afghanistan*

Institute of Medicine, Board on the Health of Select Populations, Committee on the Long-Term Health Consequences of Exposure to Burn Pits in Iraq and Afghanistan, 2011-12-01 Many veterans returning from the conflicts in Iraq and Afghanistan have health problems they believe are related to their exposure to the smoke from the burning of waste in open air burn pits on military bases Particular controversy surrounds the burn pit used to dispose of solid waste at Joint Base Balad in Iraq which burned up to 200 tons of waste per day in 2007 The Department of Veterans Affairs asked the IOM to form a committee to determine the long term health effects from exposure to these burn pits Insufficient evidence prevented the IOM committee from developing firm conclusions This report therefore recommends that along with more efficient data gathering methods a study be conducted that would evaluate the health status of service members from their time of deployment over many years to determine their incidence of chronic diseases

**Maternal alcohol use and effects on the fetus and the neonate, including the fetal alcohol syndrome (FAS)** Charlotte Kenton, 1978

**Current Bibliography of Epidemiology**, 1976 Monthly with annual cumulations Comprehensive current index to periodical medical literature intended for use of practitioners investigators and other workers in community medicine who are concerned with the etiology prevention and control of disease Citations are derived from MEDLARS tapes for Index Medicus of corresponding date Arrangement by 2 sections i e Selected subject headings and Diseases organisms vaccines No author index

*The Costs and Effects of Chronic Exposure to Low-level Pollutants in the Environment* United States. Congress. House. Committee on Science and Technology. Subcommittee on the Environment and the Atmosphere, 1976

**Risk Assessment of Prenatally-Induced Adverse Health Effects** Diether Neubert, Robert J. Kavlock, Hans-Joachim Merker, Jane Klein, 2012-12-06 Since the thalidomide Contergan tragedy about 30 years ago the induction of prenatally induced morphological or functional defects has been an area of extensive research Risk assessment of prenatally induced adverse health effects is still a difficult task from both experimental data as well as from observations in humans In the contributions to this book three major aspects are dealt with Quantitative extrapolations of experimental data to the situation possibly relevant for man The significance for a risk assessment with respect to man of minor or rare structural abnormalities observed in experimental studies The future need to assess congenital dysfunctions e g of the hormone or the immune system beside the present evaluation of structural defects Limitations as well as gaps of the present knowledge in this area of basic and applied research are pointed out Since the results of prenatally induced lesions may manifest themselves not only pre but often not before late postnatally numerous aspects of structural and functional abnormal development must be studied in experimental and clinical investigations

**Birth Defects** Janet V. Engels, 2006 Birth defects are defined as abnormalities of structure function or body metabolism that are present at birth These abnormalities lead to mental or physical disabilities or are fatal There are more than 4 000 different known birth defects ranging from minor to serious and although many of them can be treated or cured they are the leading cause of death in the first year of life This book presents leading research in this

field from around the world      **The Health Consequences of Involuntary Exposure to Tobacco Smoke** ,2006 This Surgeon General's report returns to the topic of the health effects of involuntary exposure to tobacco smoke The last comprehensive review of this evidence by the Department of Health and Human Services DHHS was in the 1986 Surgeon General's report *The Health Consequences of Involuntary Smoking* published 20 years ago this year This new report updates the evidence of the harmful effects of involuntary exposure to tobacco smoke This large body of research findings is captured in an accompanying dynamic database that profiles key epidemiologic findings and allows the evidence on health effects of exposure to tobacco smoke to be synthesized and updated following the format of the 2004 report *The Health Consequences of Smoking* The database enables users to explore the data and studies supporting the conclusions in the report The database is available on the Web site of the Centers for Disease Control and Prevention CDC at <http://www.cdc.gov/tobacco>

**Cumulated Index Medicus** ,1976      **Birth Defects** Sally Kelly,1976      **Food and Nutrition at Risk in America** Sari Edelstein,2009 This text offers readers the opportunity to consider the current status of food insecurity biotechnology food safety and bioterrorism in America as well as the types of assistance and policies needed to ensure health and welfare

Setting Priorities, Getting Results National Academy of Public Administration,1995      Moss & Adams' Heart Disease in Infants, Children, and Adolescents Hugh D. Allen,David J. Driscoll,Robert E. Shaddy,Timothy F. Feltes,2013-05-30 This 8th Edition of Moss and Adams Heart Disease in Infants Children and Adolescents Including the Fetus and Young Adult provides updated and useful information from leading experts in pediatric cardiology Added chapters and a companion web site that includes the full text with bonus question and answer sections make this Moss and Adams edition a valuable resource for those who care for infants children adolescents young adults and fetuses with heart disease Features Access to online questions similar to those on the pediatric cardiology board examination to prepare you for certification or recertification Leading international experts provide state of the art diagnostic and interventional techniques to keep you abreast of the latest advances in treatment of young patients Chapters on quality of life quality and safety pharmacology and research design add to this well respected text

Folate in Health and Disease Lynn B. Bailey,2009-11-24 During the fifteen years since the bestselling first edition of *Folate in Health and Disease* was published there have been thousands of new research studies related to folate and its role in health and disease The second edition of the book uniquely bridges the gap between basic science and public health clinical medicine Presents Groundbreaking      *Pregnancy and Birth Outcomes* Wei Wu,2018-06-13 More than 10% of the global burden of disease is due to pregnancy complications and adverse birth outcomes Despite recent advances in obstetric medicine pregnancy complications and adverse birth outcomes are a growing public health concern and economic burden on the healthcare system This book will focus on the pregnancy complications and birth outcomes from the aspects of gestational age environmental genetic epigenetic risk factors and delivery room management

Avery's Neonatology Mhairi G. MacDonald,Mary M. K. Seshia,Martha D. Mullett,2005 Revised and updated by a new

editorial team the Sixth Edition of this text will remain the leading reference on the clinical care of the newborn. It provides complete authoritative information on the biology of the newborn and the pathophysiology and management of diseases during the neonatal period. This edition has five new chapters on the role of telemedicine in neonatology, the impact of labor and delivery on the fetus, fetal determinants of adult disease, breast feeding, and control of breathing and apnea. Also included is a full color insert illustrating key signs and symptoms, selected imaging techniques, and dermatologic conditions.

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