

# Journal of Vibration Engineering

ISSN:1004-4523

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## <u>CONGENITAL ANOMALIES</u> – A REVIEW OF ENVIRONMENTAL EFFECTS ON CONGENITAL DEFORMITIES

Nikita khatri, Monika thakur, Gaurav k Singh#, Navjot kaur Department of forensic science and toxicology, university institute of applied health sciences, ChandigarhUniversity, Gharaun mohali, Punjab

(#) Corresponding Author, E-mail- gauravkumar.uips@cumail.in.

**ABSTRACT:** This paper reviews the published work on the effect of environment that are the external factors affecting the occurrence of congenital anomalies in human population. The impact of environment is hazardous on various levels in humans like the maternal exposure to pesticides, smoke, chemicals, alcohol, waste etc causes birth defects in offspring's, such defects include or facial cleft, neural tube defects, conotruncal defects, septal defects, valve defects in heart, and TDS (Testicular dysgenesis syndrome), renal dysplasia etc.

CDH(Congenital diaphragmatic hernia), CHD(congenital heart defects), right sided obstructive defects, anorectal atresia caused due to exposure to smoke by the parent.). Thus, it is said that the environment factors do play an important role in causing these birth defects or congenital anomalies in humans. A precautionary approach should be adopted at both community and individual level. In order to prevent congenital anomalies, one must reduce exposure to potential teratogens before pregnancy is recognized (i.e. preconceptionally and in the first few weeks of pregnancy).

#### INTRODUCTION:

The structural and functional defects that occurs in the intrauterine life or at the time of birth are known as congenital deformities or anomalies.

These are also termed as birth defects.

These anomalies can be categorized into 2 types:

- 1.Major body defects(MaBD)
- 2.Minor body defects(MiBD)

The congenital anomalies can also be categorised based on the types of area which they have affected.

- 1.Isolated condition These divided on the basis of anatomical locations like duplication, hypoplasia, agenisis of upper limb bone.
- 2.Generalised condition Various rare defects present at birth which affects the skeleton . Some common condition included like Acondroplasia, tumor like dysplasia and ,
- 3. Other deformities like neuromuscular or trauma or tumors, cleft lips and cleft palate etc.

- 1. The impact of environmental pollution on congenital anomalies: This paper contains epidemiological studies that have specifically looked at congenital anomalies as a possible outcome of community exposure to chemical exposures associated with environmental pollution. These include studies of drinking water contaminants (heavy metals and nitrates, chlorinated and aromatic solvents, and chlorination by-products), residence near waste disposal sites and contaminated land, pesticide exposure in agricultural areas, air pollution and industrial pollution sources, food contamination, and disasters involving accidental, negligent or deliberate chemical releases of great magnitude.
- 2. Gestational effects of maternal hyperthermia due to febrile illnesses and resultant patterns of defects in humans: Animal studies have demonstrated heat to be a significant cause for reproductive problems in a wide variety of mammals. These problems range from embryonic death and abortion to teratogenically induced anomalies, and are heavily dependent on the dose and timing of the exposure. The threshold of effect in many species begins at about 1.5 C over normal core body temperature.
- 3. Testicular dysgenesis syndrome: an increasingly common developmental disorder with environmental aspects: This article summarizes existing evidence supporting a new concept that poor semen quality, testis cancer, undescended testis and hypospadias are symptoms of one underlying entity, the testicular dysgenesis syndrome (TDS), which may be increasingly common due to adverse environmental influences. Experimental and epidemiological studies suggest that TDS is a result of disruption of embryonal programming and gonadal development during fetal life.
- 4. The risk of adverse reproductive and developmental disorders due to occupational pesticide exposure: The epidemiological studies presented in this paper refer to the association between agricultural occupation of parents and the incidence of infertility, congenital malformations, miscarriage, low birth weight, small-for-gestational-age (SGA) birth, preterm delivery and stillbirth. The results of the analyses showed that employment in agriculture increases the risk of specific morphological abnormalities in sperm, including the decreased sperm count per ejaculate and declined percentage of viable sperm. In general, no effect of exposure to pesticides on sexual hormones was observed. The data on the effect of employment in agriculture on the time to pregnancy are unequivocal, but most of them suggest that there is a relationship between the decreased fecundability ratio and pesticide exposure. Nor does the research on the sex ratio of offspring provide explicit results. The analyses indicate that parental employment in agriculture could increase the risk of congenital malformations in the offspring's, particularly such as orofacial cleft, birthmarks in the form of hemangioma as well as musculoskeletal and nervous system defects.

- 5. Maternal pesticide exposure from multiple sources and selected congenital anomalies: This paper is about the relation between various potential sources of maternal periconceptional pregnancy exposures to pesticides and congenital anomalies in offspring. Data were derived from a case-control study of fetuses and live born infants with or facial clefts, neural tube defects, conotruncal defects, or limb anomalies, among California births and fetal deaths. The orofacial cleft cases, neural tube defect cases, conotruncal defect cases, limb cases, and normal formed controls. The odds ratio (OR) estimates did not indicate increased risk for any of the studied anomaly groups among women whose self-reported occupational tasks were considered by an industrial hygienist likely to involve pesticide exposures. Paternal occupational exposure to pesticides, as reported by the mother, revealed elevated ORs for only two of the cleft phenotypes confidence interval for multiple cleft lip with/without cleft palate and for multiple cleft. Use of pesticide products for the control of pests in or around homes was not associated with elevated risks for most of the studied anomalies, although women who reported that a professional applied pesticides to their homes had increased risks for neural tube defectaffected pregnancies and limb anomalies. Women who reported living within few miles of an agricultural crop revealed increased risks for offspring with neural tube defects.
- 6. Maternal pesticide exposure and pregnancy outcome: Exposure to pesticides is inherent in many agricultural jobs. Most of the interest in connection with pesticides and pregnancy outcome has been directed to birth defects. Some indications of an elevated risk of limb anomalies have been associated with ecologic exposure, maternal environmental exposure to pesticides determined by the mother's place of residence, and parental occupation involving potential pesticide exposure. Orofacial clefts have been related to maternal environmental exposure to pesticides and exposure in agricultural work. Also, there is evidence that maternal agricultural occupation and pesticide exposure may be associated with elevated risk of spontaneous abortion and stillbirth. But, some studies have found no indication of reproductive hazards but, altogether, the epidemiologic evidence is inconclusive as regards the risk of adverse pregnancy outcome.
- 7. Maternal occupation in agriculture and risk of limb defects in Washington State, 1980—1993: This study examined the association between maternal occupational exposure to agricultural chemicals and the risk of limb defects among offspring. An elevated risk of limb defects was observed for the exposed group in comparison with both the nonagricultural and paternal agriculture groups. The results support the hypothesis that maternal occupational exposure to agricultural chemicals may increase the risk of giving birth to a child with limb defects.
- 8. Maternal residential proximity to hazardous waste sites and risk for selected congenital malformations: The investigations of whether maternal residential proximity to hazardous

waste sites increased the risk for neural tube defects, conotruncal heart defects, and oral cleft defects in California. The identification of the locations of some inactive hazardous waste sites and systematically collected information on site-related contamination for the subset of some National Priority List sites. After controlling for several potential confounders, it was found little or no increased risk for maternal residence in a census tract containing a site. Furthermore, elevated Odds were observed for neural tube defects and heart defects in association with maternal residence within 1 mile of National Priority List sites containing selected chemical contaminants.

- 9. Maternal residential exposure to hazardous wastes and risk of central nervous system and musculoskeletal birth defects: The authors used a case-control design to evaluate the risk of central nervous system and musculoskeletal birth defects relative to exposure to solvents, metal, and pesticide contaminants from hazardous waste sites. With respect to central nervous system defects, there was an elevated risk associated with living near industrial facilities that emitted solvents into the air. The low proportion of individuals who had a medium or high probability of residential exposure to hazardous waste-site contaminants limited the investigation of particular pathways, disease subgroups, and/or geographic areas. Associations between central nervous system defects and industrial releases of solvents and metals need to be investigated further
- 10. Congenital malformations and birthweight in areas with potential environmental contamination: The authors sought to determine if there was an association between a child's congenital malformation or a child's lowered weight at birth and his or her mother's residence in a census tract where a site of environmental contamination had been documented. Exposure designations were derived from existing sources of information. Except for an elevated risk for infants with malformations of the heart and circulatory system, this investigation did not reveal increased risks for most malformations or for lowered birthweight among babies born to women who lived in these census tracts.
- 11. Ambient air pollution and cardiovascular malformations in Atlanta, Georgia, 1986–2003 Temporal associations between the pollutants and daily risks of secundum atrial septal defect, aortic coarctation, hypoplastic left heart syndrome, patent ductusarteriosus, valvar pulmonary stenosis, tetralogy of Fallot, transposition of the great arteries, muscular ventricular septal defect, perimembranous ventricular septal defect, conotruncal defects, left ventricular outflow tract defect, and right ventricular outflow defect were modeled by using Poisson generalized linear models.
- 12. Ambient air pollution and congenital heart disease: Maternal exposure to ambient air pollution has increasingly been linked to adverse pregnancy outcomes. The evidence linking this exposure to congenital anomalies is still limited and

controversial. Maternal exposure to ambient air pollution has increasingly been linked to adverse pregnancy outcomes. The evidence linking this exposure to congenital anomalies is still limited and controversial.

- 13. Relation between Ambient Air Quality and Selected Birth Defects, Seven County Study, Texas, 1997–2000: There were inverse associations between carbon monoxide and isolated atrial septal defects and between ozone and isolated ventricular septal defects. Evidence that air pollution exposure influences the risk of oral clefts was limited. Suggestive results support a previously reported finding of an association between ozone exposure and pulmonary artery and valve defects.
- 14. Environmental Impacts on Congenital Anomalies-Information for the Non-Expert Professional a (limited) number of pollutants (e.g. lead, methyl mercury) well known teratogens, but also that the number of daily environmental exposures associated with congenital anomalies is increasing. This latter applies, among others, to lead and nitrates in drinking water, living near waste deposit sites and non- occupational exposure to pesticides.
- 15. The increasing incidence of hypospadias and cryptorchidism in a number of industrialised countries is noticeable. The "testicular dysgenesis syndrome" (TDS) theory links the epidemiological data with environmental causes and hypothesises one unifying mechanism for which the experimental evidence is significant.
- 16. Risk of congenital anomalies in the vicinity of municipal solid waste incinerators. The rate of congenital anomalies was not significantly higher in exposed compared with unexposed communities. Some subgroups of major anomalies, specifically facial clefts and renal dysplasia, were more frequent in the exposed communities. Among exposed communities, a dose-response trend of risk with increasing exposure was observed for obstructive uropathies. Risks of cardiac anomalies, obstructive uropathies, and skin anomalies increased linearly with road traffic density.
- 17. Maternal exposure to tobacco smoke, alcohol and caffeine, and risk of anorectal atresia: National Birth Defects Prevention Study 1997–2003: Anorectal atresia is a congenital anomaly with mostly unknown risk factors. Studies have provided evidence of teratogenic effects of alcohol and tobacco, and animal studies have suggested that caffeine may potentiate their teratogenicity. However, it is unclear how these factors affect the risk of anorectal atresia.
- 18. Prenatal and postnatal environmental tobacco smoke exposure and children's health: Children's exposure to tobacco constituents during fetal development and via environmental tobacco smoke (ETS) exposure is perhaps the most ubiquitous and

hazardous of children's environmental exposures. A large literature links both prenatal maternal smoking and children's ETS exposure to decreased lung growth and increased rates of respiratory tract infections, otitis media, and childhood asthma, with the severity of these problems increasing with increased exposure. Sudden infant death syndrome, behavioral problems, neurocognitive decrements, and increased rates of adolescent smoking also are associated with such exposures. Studies of each of these problems suggest independent effects of both pre- and postnatal exposure for each, with the respiratory risk associated with parental smoking seeming to be greatest during fetal development and the first several years of life.

- 19. Maternal smoking and congenital heart defects: Maternal smoking during pregnancy was associated with septal and right-sided obstructive defects. Additional investigation into the timing of tobacco exposure and genetic susceptibilities that could modify this risk will provide a more precise evidence base on which to build clinical and public health primary prevention strategies.
- 20. Maternal Alcohol Use in Relation to Selected Birth Defects: The hypothesis that maternal alcohol consumption affects the development of structures possibly derived from a common embryonic cell population, the cranial neural crest, was explored using data collected by a case-control surveillance program of birth defects. Maternal alcohol use is less related to overall malformations derived from cranial neural crest cell than to one specific defect among them—cleft lip with or without cleft palate.
- 21. Maternal periconceptional folic acid intake and risk of autism spectrum disorders and developmental delay in the CHARGE (CHildhood Autism Risks from Genetics and Environment) case-control study. According to this paper Periconceptional folic acid may reduce ASD risk in those with inefficient folate metabolism. Maternal periconceptional exposure to cigarette smoking and alcohol consumption and congenital diaphragmatic hernia.
- 22. Congenital diaphragmatic hernia (CDH) is a major birth defect that occurs when abdominal organs herniate through a diaphragmatic opening into the thoracic cavity and is associated with high mortality (> 50%): These findings identified periconceptional smoking exposure as a potential risk factor for CDH. Future studies need to confirm our findings and explore possible pathways accounting for the teratogenic effect of smoking. Maternal periconceptional use of electric bed-heating devices and risk for neural tube defects and orofacial clefts

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23. Electric and magnetic fields are of concern as risk factors for adverse reproductive outcomes, including congenital anomalies. Among residential exposures to electric and magnetic fields, electric bed-heating devices such as electric blankets may be a substantial source of such exposures, and their use is fairly common. Two population-based case-control studies were analyzed to investigate whether the periconceptional use of electric blankets, bed warmers, or electrically heated waterbeds increased the risk of women to deliver infants or fetuses with neural tube defects (NTDs) or orofacial clefts.

#### **CONCLUSION:**

This paper compiles the different environment that are external factors responsibile for causing congenital anomalies in human population. These factors include exposure to smoke, alcohol by the mother or the residency near the hazards site, landfills or sewage treatment plants or dumped industrial waste and agriculture farms etc. The common anomalies found due to above mentioned factors can be categorized as lung related like asthma, heart related like congenital heart defects, valve defects, septal defects, nervous system related like neural tube defect, affect on Central nervous system, kidney related like renal dysplasia, the muscular skeleton system and some physical abnormalities like orofacial cleft lip, cleft palate, and some reproductive organ defects like TDS (testicular dysgenesis syndrome). Thus, it is concluded that the environment factors do play an important role in causing these birth deformities or congenital anomalies in humans. A precautionary approach should be adopted at both community and individual level. In order to prevent congenital anomalies, one must reduce exposure to potential teratogens before pregnancy is recognized (i.e. preconceptionally and in the first few weeks of pregnancy). It is also concluded that there are relatively few environmental pollution exposures for which we can draw strong conclusions about the potential to cause congenital anomalies and, if so, the chemical constituents implicated, to provide an evidence base for public health and clinical practice.

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