Background and Aims: Maternal exposure to environmental chemical mutagens (heavy metals, formaldehyde) may enhance risk for congenital malformations and chromosome aberrations in fetuses (newborns). We aimed to determine cytogenetic effect markers in mothers and fetuses exposed to environmental chemical mutagens using epidemiological study data.

Methods: A cytogenetic examination of 95 mother-newborn pairs exposed to environmental chemical mutagens including manganese, nickel, lead, formaldehyde (a study group), and 70 non-exposed dyads (a control group) was carried out analyzing metaphase chromosomes from mother and newborn’s blood lymphocytes by the standard method. Blood chemical mutagen concentrations were determined applying atomic absorption spectrophotometry, liquid chromatography. The relationship between the frequency of chromosome aberrations, and the exposure to the chemical mutagens was assessed using epidemiological study data and calculating odds ratio (OR).

Results: The blood samples from the pregnant and parturient women were found to contain elevated formaldehyde, manganese, nickel, lead concentrations (1.5-12 fold increase compared to the reference levels) (p=0.001). Polymorphic chromosome changes were detected in 30% of the study group women that exceeded the control group indices by 5-fold. The probability (OR) of chromosome aberrations in newborns from the exposed women with chromosome polymorphism was higher (OR=6.7) compared to the control group. We determined the probability of chromosome aberrations in newborns whose mothers demonstrated high frequency of chromosome polymorphism associated with the exposure to formaldehyde and nickel (OR=10.5, p=0.001). The probability of numerous congenital malformations and chromosome aberrations was higher in newborns of women exposed to manganese, lead and nickel having an increased frequency of chromosome polymorphism (OR=2.7, p=0.001).

Conclusions: Exposure markers are elevated concentrations of manganese, formaldehyde, nickel, lead in the mothers and newborns’ blood. Cytogenetic effect markers are high frequency of chromosome polymorphism in the mothers and associated chromosome aberrations in the newborns.