AB015. Study on pericentric inversion of chromosome 9 and congenital abnormalities in children

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Background: Pericentric inversion of chromosome 9 [inv(9)] is one of the most common human chromosomal variations. The aim of this study was to detect inv(9) from karyotype in children and its clinical correlation with birth defects.

Methods: The chromosome analysis using G-banding was carried out in 8,869 suspected children with congenital abnormalities at Children's Hospital Central from 2013 to May 2017. All of their clinical data will be reviewed.

Results: Out of 8,869 cases, 2,361 chromosomal aberration cases were detected (26.6%). Chromosomal polymorphisms were detected in 267 patients (3%). In 267 patients with chromosomal polymorphisms, the most common of chromosome variation was inv(9), found in 60 patients (22.5%). Nine patients with inv(9) have other chromosomal abnormalities. Various congenital anomalies in patients with inv(9) consist of malformation, dysmorphic face, cerebral palsy, congenital heart defect, esophageal shrinkage, urogenital anomalies, inguinal hernia, hypospadias, and malformed anus.

Conclusions: The inv(9) has been regarded as a normal variant in human population without phenotypic consequence. However, inv(9) would be expected to be associated with congenital anomalies in children. Many reports indicated that inv(9) has been associated with infertility, recurrent pregnancy loss. Few studies suspected that inv(9) is related to cancer such as ovarian cancers, acute leukemia, chronic myeloid leukemia. The parental chromosome analysis and genetic counseling are essential for children with inv(9).

Keywords: Pericentric inversion; chromosome 9; inv(9); congenital abnormalities

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