INCIDENCE OF TRISOMY 18 PRENATALLY DIAGNOSED BY AMNIOCENTESIS - ONE CENTER EXPERIENCE

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Abstract: The Trisomy 18 Syndrome (Edwards Syndrome) occurs due presence of an extra chromosome 18 (full or mosaic trisomy) or partial trisomy 18q and it is the second most common autosomal trisomy after trisomy 21. The aim of this study was to determine the incidence of Edwards Syndrome in the population of fetuses whose mothers underwent diagnostic amniocentesis for fetal karyotyping and display characteristics of individual cases. We performed a retrospective analysis of data on 5421 pregnant women who made amniocentesis over 11 years. The incidence of fetuses with trisomy 18 estimated as 1: 493 in pregnancies that were followed by amniocentesis (0.2%). It is noted significantly more (P<0.5) male fetuses with Edwards Syndrome compared to female (72.7% vs. 27.3%). These findings are inconsistent with the clinical reports that showed higher prevalence at birth in female.

Keywords: trisomy 18, Edwards Syndrome, amniocentesis