OC04.05

Trisomy 18 and Trisomy 13 in Denmark from 1997-2007; impact of a national change in screening strategy for Trisomy 21

C.K. Ekelund[1], L. Skibsted[2], O.B. Petersen[3], A. Tabor[1]
[1]Department of Fetal Medicine, Rigshospitalet, Copenhagen, Denmark; [2]Deparment of Fetal Medicine, Roskilde Sygehus, Roskilde, Denmark; [3]Department of Fetal Medicine, Aarhus Universitetshospital Skejby, Aarhus, Denmark

Objectives: To evaluate number of pre- and postnatally detected cases of Trisomy 18 and Trisomy 13 in relation to the introduction of a new national screening strategy for Trisomy 21 issued in 2004.
Methods: All cases of Trisomy 18 and Trisomy 13 detected pre- and postnatally from 1997 to 2007 were
retrieved from the National Danish Cytogenetic Registry including information on gestational age at diagnosis for prenatal cases.
Results: The number of cases of Trisomy 13 and Trisomy 18 detected after 22 weeks or at birth has decreased from 15-20 cases per year in 1997-2004 to less than 5 cases in 2007 (figure 1). The proportion of prenatal cases detected before 18 weeks has increased after 2004.
Conclusion: Introduction of new national guidelines, offering a combined first trimester risk assessment for Trisomy 21 to all pregnant women, have also had an impact on the detection of other chromosomal aneuploidies. The vast majority of fetuses with Trisomy 13 and Trisomy 18 are diagnosed early in pregnancy, even though the number of invasive procedures performed in Denmark after the introduction of the new national guidelines has decreased steeply.

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Figure 1