

Perinatal diagnosis of chromosomal aneuploidies

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Objective

To determine the prevalence of circumstances of diagnosis of chromosomal aneuploidies.

Methods

We conducted a retrospective study in the department of neonatology of the Charles Nicolle Hospital of Tunis, over a 3 years period. We included all cases of chromosomal aneuploidies.

Results

During the study period, 50 cases of chromosomal aneuploidies were diagnosed with an incidence of 4, 3‰ births. The aneuploidies most commonly found were Trisomy 21, 66% (33/50) of the cases, T18 in 18% of cases (9/50) and T13 in 10% of cases (5/50). Chromosomal aneuploidies were isolated in 72% of cases (36/50) and associated with major birth defects in 28% (14/50). The perinatal rate mortality was 32% (16/50).

Conclusion

Our study showed a high mortality rate because of a lack of antenatal diagnosis.