

Trisomy 13 and 18: the Comfort Care option

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Objective. Trisomy 13 and 18 are chromosomal abnormalities associated with limited life expectancy. Diagnosis can be suspected through first-trimester screenings (combined test and foetal DNA testing) or after detecting foetal malformations via obstetric ultrasounds, and confirmed by foetal karyotype analysis. For couples who choose to continue the pregnancy, perinatal palliative care (PPC) is an option. The goal of PPC is to focus not only on the foetus/child but also on the well-being of the entire family, aiming to minimize psychological trauma. This study aims to describe cases of trisomy 13 and 18 diagnosed at our prenatal diagnosis centre.

Materials and Methods. We conducted a retrospective study on pregnancies diagnosed with trisomy 13 and 18 at our hospital from 2010 to 2023. All patients received personalized

multidisciplinary counselling on the clinical picture and available care options, including PPC. Cases of chromosomal mosaicism were excluded.

Results. During the study period, we diagnosed 60 cases of trisomy 13 (n = 13, 22%) and trisomy 18 (n = 47, 78%). Of these, 22 women (37%) decided to terminate the pregnancy; 3 pregnancies (5%) resulted in miscarriage; 3 women (5%) were lost to follow-up; and 32 women (53%) chose to continue with PPC. Among these, 15 experienced intrauterine foetal death. Of the 14 live foetuses at the onset of labour, 3 were stillborn, and 11 had survival times ranging from under 1 hour to 3 years.

Conclusions. A significant number of couples opted for PPC. Prenatal diagnosis centres should offer personalised care and the PPC option in collaboration with neonatologists.