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Antenatal diagnosis of trisomy 18, harm and parental choice

Dominic J C Wilkinson

In this commentary I assess the possible harms to a fetus with trisomy 18 of continued life. I argue that, although there is good reason to avoid subjecting infants to major surgery and prolonged intensive care where there is little chance of benefit, doctors should support and engage honestly with parents who decide to continue their pregnancies. We should ensure that infants with trisomy 18 have access to high quality palliative care.

In this issue, Pauline Thiele provides a moving and disturbing account of medical abandonment.¹ Following the prenatal diagnosis of a severe chromosomal abnormality (trisomy 18), she and her husband declined the option of termination of pregnancy. They then encountered apathy, misunderstanding, miscommunication and apparent hostility from medical professionals.¹ Why was she treated in this way? We could understand (though we may or may not agree with) those who object to decisions to terminate a pregnancy, perhaps because of beliefs about the moral status of human fetuses. But what reasons could there be for criticising or condemning mothers who choose *not* to terminate a pregnancy where a fetus is affected by a severe abnormality?

Trisomy 18 is the second most common autosomal trisomy (after trisomy 21) and is present in 1 out of 2600 pregnancies.² It is associated with congenital abnormalities of various organs including the heart, kidney, gastrointestinal tract, spinal cord and brain.³ The overall prevalence of trisomy 18 is increasing because of increasing maternal age although, at the same time, the birth prevalence is falling because of prenatal testing.² In many countries the majority of mothers who have an antenatal diagnosis of trisomy 18 decide to terminate their pregnancy.²

Trisomy 18 used to be classified as a 'lethal congenital abnormality'.⁴ One widely cited population study from the early 1990s found that infants born with trisomy 18 had a median survival of just 3 days, and no infant survived for more than 1 year.⁵ Thiele was told bluntly that her child was not going to live, and this is similar to the accounts of other parents who were told by professionals that

trisomy 18 was 'incompatible with life'.⁶ If it were the case that trisomy 18 led inevitably to fetal death in utero or to early neonatal death, it would be understandable were many parents to choose to terminate their pregnancy and face the death sooner rather than later. But why should mothers who elect to continue their pregnancy be subject to disapprobation? There is no good reason to think that fetuses with trisomy 18 suffer in utero. With good palliative care it should be possible to ensure that an infant who dies in the newborn period is kept comfortable and pain-free.⁷

In any case, early death is not inevitable for fetuses and infants with trisomy 18. High mortality figures in previous studies were likely influenced significantly by self-fulfilling prophecies,⁸ and by infants not receiving treatment for apnoea or for their congenital heart disease.⁴ Studies from Japan, where infants with trisomy 18 receive respiratory support if required and surgery for congenital anomalies, have shown median survival of 150–250 days, with 25–44% of children surviving for more than 1 year.^{9 10}

Is life with trisomy 18 not worth living? Some have argued that it is contrary to the best interests of infants with trisomy 18 to resuscitate them because 'they have no reasonable hope of benefit'.¹¹ If that were the case, then it might be a harm to a fetus to continue a pregnancy to full term. However, a small proportion of children with trisomy 18 survive to middle or late childhood or even adulthood.¹² They have severe or profound developmental delay and later intellectual impairment, but older children are able to ambulate, understand words and phrases and interact with those around them.^{13 14} Although severe cognitive impairment may reduce the benefits available to them,¹⁵ these children and their families do appear to gain from their life. We do not think that non-human animals with equivalent cognitive capacities are harmed by their

existence, nor that it would be better for them if they had not existed. One legitimate concern might be about the pain caused to infants by major cardiac or other surgery and prolonged stay in intensive care. For infants with trisomy 18 who have severe physical abnormalities, the burden of treatment may outweigh the reduced benefit if they survive.¹⁵ But in a recent survey of parents of children with trisomy 18 who had visited online support websites, only a small proportion of newborn infants had required mechanical ventilation; most did not have prolonged stay in hospital in the newborn period and none received major surgery.¹⁴ This may not be a representative sample, but not all infants with trisomy 18 require burdensome medical interventions.

Attitudes towards trisomy 18 are changing.¹¹ The availability of information, stories and pictures on the internet encourages some parents to continue their pregnancies and to request treatment that previously would not have been offered. Although medical professionals have been guilty (and, from Thiele's account, still are guilty) of presenting an overly negative picture of prognosis for fetuses with trisomy 18, there is also a danger that information on the internet provides an overly positive and unrealistic impression. Even with full treatment, a large proportion of infants born with trisomy 18 die in the first year of life.¹⁰ Survivors are severely or profoundly impaired.¹³

There is good reason to make sure that infants with trisomy 18 do not suffer unnecessarily and that they are not subjected to major surgery or prolonged intensive care where there is little chance of benefit. But there is no reason to treat families the way that Pauline Thiele and her husband were treated. We should support and engage with parents who choose to continue their pregnancy, we should be honest with them about treatment options and outcomes, and we should provide high quality palliative care. In that way we can help families to make the time that they have with their children as good as possible.

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