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Review article

Fatally flawed? A review and ethical analysis of lethal congenital malformations

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Prenatally diagnosed abnormalities that are associated with death in the newborn period are often referred to as 'lethal malformations'. Yet, for many of the commonly described lethal malformations long-term survival is possible if supportive interventions are provided. In this paper we analyse and review fetal or congenital lethal abnormalities. The designation 'lethal' overlaps with the concept of 'medical futility'. The term is used for a heterogenous group of conditions, and hinders clear communication and counselling. We argue that the term should be avoided, and propose in its place a set of key questions that should be addressed by counselling.

Keywords Ethics, fatal outcome, prenatal diagnosis.

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Introduction

Antenatal diagnosis of a severe congenital malformation is associated with profound grief, sadness and anger.[1–3] In many parts of the world diagnoses of this nature are often followed by decisions to terminate a pregnancy, particularly where the condition is associated with a high rate of prenatal (or stillbirth) or neonatal death.[4–7] Those conditions with the poorest prognosis are often labelled 'lethal'. This term is sometimes used in perinatal practice to convey the gravity of the fetus' condition, and may permit different obstetric decision-making.[8]

In this paper we review and analyse the concept of 'lethality' as applied to fetal or congenital malformations. The term is used in a way that is similar to 'futile treatment' in end-of-life decisions.[9] It is used for a heterogenous group of conditions to imply an ethical conclusion rather than to represent a clear prognosis: it obscures rather than aids communication and counselling. We argue that the term should be avoided, and propose an alternative framework for counselling.

As a background to this non-systematic review we searched Medline using the keywords 'lethal anomaly' 'lethal anomalies' or 'lethal malformation', or the combination of 'lethal' with the Medical Subject Headings (MeSHs) 'prenatal diagnosis' or 'congenital anomalies/diagnosis'. Cohort studies, case series, reviews or commentaries that discussed diagnosis, counselling or management for this group of conditions were identified and retrieved. Additional articles were identified from the reference lists of identified papers. Papers were searched for definitions or lists of lethal malformations. For the malformations most frequently included in lists, a separate search strategy used the condition name in combination with 'outcome' or 'survival', and identified cohort studies, case series or case reports reporting duration and frequency of survival beyond the newborn period.

What are lethal malformations?

Our search of the published literature on lethal malformations yielded 1717 papers, of which 75 full-text papers were reviewed. However, closer study revealed no agreed definition of a 'lethal fetal or congenital malformation' (LM), nor could we identify an agreed list of conditions that fit into this category.

Conventionally, 'lethal' is used to describe something (e.g. an action or agent) that will cause death.[10] There are several different ways of theoretically interpreting the description of a perinatal condition as being lethal.

1 A condition that invariably leads to fetal death *in utero*.
2 A condition that invariably leads to death *in utero* or in the newborn period, regardless of attempted supportive treatment.
3 A condition that usually leads to death *in utero* or in the newborn period.
4 A condition that has been associated with death *in utero* or in the newborn period.

Which of these definitions is the correct one?

The most straightforward understanding of LM is probably the second in the list above: a condition incompatible with survival beyond the newborn period. This terminology features in a number of reports.[1,7,11–13] Women who have been counselled following the diagnosis of trisomy 13 and 18 report that phrases like 'incompatible with life' are used by clinicians.[13–15] However, as will be discussed in the next section, definition 2 does not accurately describe any of the congenital malformations that are commonly described as being lethal. (The first definition listed above also fails to fit any of the commonly listed conditions, and we have not found it used in any of the papers that we reviewed.) Some papers use the third definition, referring to malformations that lead to death or stillbirth in a high proportion (>50, >85 or >90%) of past cases.[16–19] Older papers that referred to LMs appeared to use the fourth definition. LM was simply used to describe conditions that had been observed to cause death in epidemiological studies of perinatal mortality.[20–22]

The diagnosis of an LM has potential ethical and legal implications. Some authors identify these malformations as triggers for the involvement of perinatal palliative care services.[12,23,24] LM may warrant different obstetric management, with interventions focused largely or solely on maternal rather than fetal wellbeing.[25] It has been argued that termination of pregnancy in the third trimester would be justifiable in the presence of a certain diagnosis of an LM, as the presence of such a malformation precludes any benefit to the fetus from the pregnancy continuing,[8] or exempts the decision from legal clauses that prevent late abortion on the grounds of viability.[26] In some parts of the world, LMs represent one of the only situations when abortion is legally permitted.[4]

Which conditions are 'lethal'? There are a large number of individually rare conditions that have received this label. Lists differ, although some conditions are more frequently cited than others (Table 1).

Table 1. Malformations cited in lists of LM (arranged in descending order of frequency of citation)[7,16,18,23–25,38,56–58]

Lethal malformations
Potter's syndrome/renal agenesis
Anencephaly/acrania
Thanatophoric dwarfism
Trisomy 13 or 18
Holoprosencephaly
Triploidy
Hydranencephaly
Some cases of hypoplastic left heart syndrome and pentalogy of Cantrell
Severe osteogenesis imperfecta
Multicystic/dysplastic kidneys
Polycystic kidney disease
Congenital severe hydrocephalus with absent or minimal brain growth
Severe congenital diaphragmatic hernia with hypoplastic lungs
Sirenomelia
Complex or severe cases of meningomyelocele
Large encephaloceles
Acardia
Some cases of giant omphalocele
Inoperable conjoined twins
Cranioradischisis
Exencephaly
Iniencephaly
Harlequin fetus
Meckel–Gruber syndrome
Non-immune hydrops with major cardiac defects

Conditions are listed as cited, we do not necessarily endorse descriptions of these conditions as 'lethal'. This list does not include extreme prematurity (for example 22 weeks of gestation) severe oligohydramios/growth restriction or marked subcutaneous bullous cysts, although sometimes these conditions are grouped with LM, and similar issues apply to them.

Are lethal malformations really lethal?

Table 2 presents a summary of published outcome data for the most commonly cited LMs. These conditions are all uncommon, although prevalence estimates are hampered by the large proportion of cases that are diagnosed antenatally and followed by termination of pregnancy.[27] Postnatal survival figures are also imprecise. Firstly, these represent only live births. They are unlikely to have identical features to the larger cohort of fetuses diagnosed antenatally. Secondly, survival is influenced by decisions made before and after birth. Where a large proportion of infants receive palliative care, this may become a self-fulfilling prophecy.[28] Recently, there have been attempts to document survival in cohorts of children with trisomy 13 or 18 or holoprosencephaly.[6,29–32] Survival rates from parent-support group surveys or from case series of referred children with these conditions report considerably longer durations of survival than previous estimates.[31,32] However, it is likely that these estimates are influenced by selection bias, and represent less-severely affected individuals.

Despite the problems with available data, none of the malformations that are most commonly described as being lethal are actually lethal in the strict sense (definitions 1 or

Table 2. Published outcome for severe congenital anomalies frequently described as lethal

	Prevalence	Median survival	Proportion surviving >1 week	Longest reported survival
Renal agenesis	1.7/10 000[59]	<24 hours[59]	None reported	None reported
Anencephaly	10/10 000 pregnancies 2.6/10 000 births[60]	<24 hours[61,62] 55 minutes[63]	3–5%[61,64]	10 months,[65] 2.5 years[44]
Thanatophoric dysplasia	0.28/10 000[66]	Not reported	Not reported	5,[67] 9 years[68]
Trisomy 13	1.2/10 000[60]	7–8 days[29,30]	58%[69] 8% >1 year[29]	19,[72] 27 years[73]
Trisomy 18	2.6/10 000[60]	6–14 days[29,30]	52%[69] 8% >1 year[29]	27,[70] 30,[40] 50 years[71]
Holoprosencephaly	0.5/10 000[74]	4–5 months[31]	5/9 in series from Hungary[74]	6,[75] 11,[31] 13,[74] 19[76] years
Triploidy	1 in 10 000[77]	Not reported	Not reported	7 months[78]
Hydranencephaly	1–2.2/10 000[79]	1 month[79]	50%,[16] 15%, >1[79]	22,[80] 32 years[81]

2 above). Prolonged survival has been described in all of the conditions listed, with the exception of bilateral renal agenesis (and even in this condition survival would be conceivable with intensive support).[33]

Lethality and futility

The concept of a lethal fetal/congenital malformation has some similarities with another concept, that of 'futile' treatment. 'Medical futility' emerged in response to a perceived increase in conflicts between doctors and patients – particularly conflicts about life-sustaining treatment.[34] The idea was that if futile treatments could be confidently identified, doctors would be ethically justified in refusing to provide them.[35] However, despite receiving a lot of attention in the ensuing 20 years, there are a number of well-described problems with the concept of futility.[36,37]

The first of these is the difficulty in defining futile treatment. At least five different definitions of futility have been proposed.[34] Sometimes futility is divided into *quantitative* and *qualitative* forms. Treatment is labelled quantitatively futile if it has a very low chance of success (for example <3%).[35] However, there is no agreement about just how low the chance of success must be for treatment to be quantitatively futile. Qualitative futility is invoked where treatment might be technically successful in sustaining life, yet it is perceived that this offers no benefit to the patient. One example might be the provision of intensive care to a patient in a persistent vegetative state.[35] However, the challenge for qualitative futility is that there is considerable disagreement about what level of cognitive function is necessary to provide benefit to the patient.

These definitional problems also apply to LM. There is no agreement on the probability of death that would justify describing a condition as 'lethal'. Similarly, whereas some definitions of LM are similar to qualitative futility in including conditions that give rise to a persistent vegetative state or absent cognitive development,[11,25,38,39] there is no agreement about just how much function would justify this label. Surviving children with holoprosencephaly, hydranencephaly, and trisomy 13 and 18 have been reported to experience awareness of those around them, hear and respond to sound, and to learn and remember.[31,40,41]

The second major problem with futility (and with lethality) is the difficulty in determining whether treatment meets a given definition. As discussed in the previous section, the problem of self-fulfilling prophecies make it difficult to know how often (if ever) survival would be possible if all treatment were provided. Studies of prognosis that purport to be able to define groups of patients in whom treatment is futile are often based on insufficient data to provide statistical confidence.[42]

The third challenge for futility is that although it is possible to define subgroups of patients who meet strict criteria for the futility of treatment, this ends up applying to a relatively narrow set of cases. For example, one proposed definition uses the concept of 'physiological futility': treatment that is unable to achieve its physiological aim.[34] Analogously, 'lethality' might be restricted to conditions that are universally associated with perinatal death, regardless of treatment (definition 2). However, of the conditions that are frequently termed 'lethal', this definition appears to apply only to renal agenesis, and may not even be strictly true in this case.

The language of lethality

To our knowledge, there are no published reports of how frequently the term 'lethal' is used in counselling following prenatal diagnosis. Some practitioners may avoid it, for the reasons given above. However, our experience, coupled with the literature summarised above and the accounts of parents, suggests that a significant proportion of practitioners do use this or related terminology. Why is the term used? One reason is that practitioners may mistakenly believe that conditions like those listed in Tables 1 and 2 are not compatible with survival beyond the newborn period. A second potential reason is a discomfort with uncertainty, or a desire to make decision-making simpler. It is

potentially easier for women to come to terms with termination of pregnancy or with palliative care if they are told that survival is impossible, and it may be easier for professionals to make and rationalise such decisions. A third possibility is that practitioners are aware that death is not inevitable but believe that the survivors will not have a life that is worth living. Finally, it may be that the term 'lethal' is believed to be an effective way of communicating the grave nature of the fetus' prognosis.

But does it matter what term is used? As long as we are clear what is meant by a set of words, it may not be important. But the problem is that 'lethal malformation' is not clear, and the way that it is used hampers, rather than helps, communication. For example, Barr and Cohen note that families of children with holoprosencephaly are often told (or gain the impression) that their children will die soon after birth. However, if the infants do not die there are major repercussions for trust between families and health professionals.[31] Other parents describe deep senses of disillusionment and anger at being misled by health professionals.[14,15,43] More significantly, perhaps, it has been argued that the term lethal is dangerous, as it contains concealed judgements about the quality of life of affected infants, and risks taking decisions out of the hands of women.[44,45]

Doctors discuss a wide range of medical decisions with patients. They provide information about medical diagnoses and treatment options. In many circumstances, doctors also provide guidance about which course of action they feel would be best. Counselling following prenatal diagnosis is distinctive, in that health professionals are exhorted to avoid making value judgements and to avoid explicit recommendations.[46–48] Although some have challenged whether complete neutrality in counselling is achievable,[49,50] or desirable,[46,51] there are nevertheless good reasons why doctors should aspire to neutrality in most circumstances. One of the important ways to enhance the autonomy of women is to use language that avoids concealed value judgements.[52]

Counselling and management

If we avoid the term 'lethal', then how should women be counselled about diagnoses such as those listed in Table 1? Rather than trying to find some new term to encompass a group of conditions that are heterogenous, our recommendation is that counselling should focus on addressing five key questions that are the basis for understanding the fetus' prognosis (Box 1). These questions will be challenging, as the answers to some of them are likely to be uncertain. Nevertheless, physicians have a professional responsibility to provide the best available evidence to address them. Acknowledging the limits of the available data and genuine uncertainties in prognosis is important if a woman's autonomy is to be respected.

Box 1. Key questions for counselling on severe congenital malformations

1 What is the diagnosis and how certain is it?
2 What is the likelihood of survival beyond the newborn period if life-sustaining treatment is provided (for example mechanical ventilation or surgery)?
3 What is the likely duration of survival if life-sustaining treatment is provided?
4 What is the range of possible physical or cognitive impairments if the newborn survives?
5 What is the burden of treatment required to keep the baby alive?

One important point to note is that changing the terminology that is used to describe or to classify severe congenital malformations does not, in itself, change the management options that could ethically be provided. For example, in our view it would be appropriate to offer couples the options of perinatal palliative care and maternal-focused obstetric management in the third trimester for all of the conditions listed in Table 2 (whether or not they are called 'lethal'). The high probability of death despite treatment, the nature of the impairments that infants would experience if they survived, and the burdens to the infant of treatment make it genuinely questionable whether life-prolonging treatments are appropriate. Where treatment would cause a child to suffer, but yield little or no benefit because he or she lacks the capacity to develop even minimal relationships with those around them,[53] it may be unethical to provide it. However, we also agree with others who have argued that it would, in some circumstances, be appropriate to provide intra-partum management that is aimed at live birth,[54] and it may sometimes be consistent with the child's best interests to provide life-sustaining treatments, including intensive care.[55]

The ethics of termination of pregnancy in the third trimester is beyond the scope of this paper. However, in countries where the diagnosis of a 'lethal malformation' is legally required prior to termination, there may need to be further clarification about which definition of LM is to be used.

Conclusions

This paper has outlined the serious conceptual and practical challenges inherent in defining 'lethal malformations'. The term 'lethal', unless clearly and transparently defined, and consistently applied, should be avoided in perinatal guidelines and in counselling following prenatal diagnosis. It runs the risk of misleading couples about the prognosis of severe abnormalities. Counsellors should be explicit about prognosis if treatment is provided, and about the implications of treatment for the fetus or infant. This will, in many cases, require the acknowledgement of uncertainty.

Disclosure of interests
The authors have no relevant financial, political, intellectual or religious interests to disclose. DW and AW work clinically in newborn intensive care. LdC works as an ultrasonologist, and counsels women following prenatal diagnosis. PT has had personal experience of a severe fetal malformation.

Contribution to authorship
DW conceived of, conducted the literature review and completed the first draft of this work. PT, AW and LdC contributed to the design of the article, ethical analysis and edited the article. All authors approved of the final version of the article.

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References

1 Hunfeld JAM, Wladimiroff JW, Passchier J, Uniken Venema-van uden M, Frets PG, Verhage F. Emotional-Reactions in Women in Late Pregnancy (24 Weeks or Longer) Following the Ultrasound Diagnosis of a Severe or Lethal Fetal Malformation. *Prenat Diagn* 1993;13:603–12.
2 Leuthner S, Bolger M, Frommelt M, Nelson R. The impact of abnormal fetal echocardiography on expectant parents' experience of pregnancy: a pilot study. *J Psychosom Obstet Gynecol* 2003;24:121–9.
3 Senanayake H, De Silva D, Premaratne S, Kulatunge M. Psychological reactions and coping strategies of Sri Lankan women carrying fetuses with lethal congenital malformations. *Ceylon Med J* 2009;51:14–7.
4 Benute GRG, Nomura RMY, Liao AW, Brizot MdL, de Lucia MCS, Zugaib M. Feelings of women regarding end-of-life decision making after ultrasound diagnosis of a lethal fetal malformation. *Midwifery* 2011; [Epub ahead of print] doi:10.1016/j.midw.2011.06.011.
5 Hassed S, Miller C, Pope S, Murphy P, Quirk J Jr, Cunniff C. Perinatal lethal conditions: the effect of diagnosis on decision making. *Obstet Gynecol* 1993;82:37–42.
6 Lakovschek IC, Streubel B, Ulm B. Natural outcome of trisomy 13, trisomy 18, and triploidy after prenatal diagnosis. *Am J Med Genet A* 2011;155:2626–33.
7 Schechtman KB, Gray DL, Baty JD, Rothman SM. Decision-making for termination of pregnancies with fetal anomalies: analysis of 53 000 pregnancies. *Obstet Gynecol* 2002;99:216–22.
8 Chervenak FA, Farley MA, Walters L, Hobbins JC, Mahoney MJ. When is termination of pregnancy during the third trimester morally justifiable? *N Engl J Med* 1984;310:501–4.
9 Sokol DK. The slipperiness of futility. *BMJ* 2009;338:b2222.
10 *The Oxford English Dictionary*. 2nd edn. OED Online: Oxford University Press, Oxford 1989.
11 World Health Organization. *Basic Newborn Resuscitation: A Practical Guide*. [online] World Health Organization, 1998.
12 Catlin A, Carter B. Creation of a neonatal end-of-life palliative care protocol. *J Perinatol* 2002;22:184–95.
13 Munson D, Leuthner SR. Palliative care for the family carrying a fetus with a life-limiting diagnosis. *Pediatr Clin North Am* 2007;54:787–98, xii.
14 Walker LV, Miller VJ, Dalton VK. The health-care experiences of families given the prenatal diagnosis of trisomy 18. *J Perinatol* 2008;28:12–9.
15 Thiele P. He was my son, not a dying baby. *J Med Ethics* 2010;36:646–7.
16 Courtwright A, Laughon M, Doron M. Length of life and treatment intensity in infants diagnosed prenatally or postnatally with congenital anomalies considered to be lethal. *J Perinatol* 2011;31:387–91.
17 Czeizel AE. First 25 years of the Hungarian congenital abnormality registry. *Teratology* 1997;55:299–305.
18 Dommergues M, Mandelbrot L, Mahieu-Caputo D, Boudjema N, Durand-Zaleski I, ICI Group-Club de médecine foetale. Termination of pregnancy following prenatal diagnosis in France: how severe are the foetal anomalies? *Prenat Diagn* 2010;30:531–9.
19 Lewis S, McGillivray G, Rowlands S, Halliday J. Perinatal outcome following suspected fetal abnormality when managed through a fetal management unit. *Prenat Diagn* 2010;30:149–55.
20 Goldenberg RL, Humphrey JL, Hale CB, Wayne JB. Lethal congenital anomalies as a cause of birth-weight-specific neonatal mortality. *JAMA* 1983;250:513–5.
21 Milunsky A. Lethal congenital anomalies. *JAMA* 1983;250:517–8.
22 Young ID, Clarke M. Lethal malformations and perinatal mortality: a 10 year review with comparison of ethnic differences. *BMJ* 1987;295:89–91.
23 Bhatia J. Palliative care in the fetus and newborn. *J Perinatol* 2006;26(Suppl 1):S24–6; discussion S31–3.
24 Breeze ACG, Lees CC, Kumar A, Missfelder-Lobos HH, Murdoch EM. Palliative care for prenatally diagnosed lethal fetal abnormality. *Arch Dis Child Fetal Neonatal Ed* 2007;92:F56–8.
25 Chervenak FA, McCullough LB. An ethically justified, clinically comprehensive management strategy for third-trimester pregnancies complicated by fetal anomalies. *Obstet Gynecol* 1990;75:311–6.
26 Strong C. Fetal anomalies: ethical and legal considerations in screening, detection, and management. *Clin Perinatol* 2003;30:113–26.
27 Irving C, Richmond S, Wren C, Longster C, Embleton ND. Changes in fetal prevalence and outcome for trisomies 13 and 18: a population-based study over 23 years. *J Matern Fetal Neonatal Med* 2011;24:137–41.
28 Wilkinson D. The self-fulfilling prophecy in intensive care. *Theor Med Bioeth* 2009;30:401–10.
29 Rasmussen SA, Wong L-YC, Yang Q, May KM, Friedman JM. Population-based analyses of mortality in trisomy 13 and trisomy 18. *Pediatrics* 2003;111:777–84.
30 Brewer CM, Holloway SH, Stone DH, Carothers AD, FitzPatrick DR. Survival in trisomy 13 and trisomy 18 cases ascertained from population based registers. *J Med Genet* 2002;39:e54.
31 Barr M, Cohen MM. Holoprosencephaly survival and performance. *Am J Med Genet A* 1999;89:116–20.
32 Bruns D. Birth history, physical characteristics, and medical conditions in long-term survivors with full trisomy 13. *Am J Med Genet A* 2011;155:2634–40.
33 Campbell N. When care cannot cure: medical problems in seriously ill babies. In: Kuhse H, Singer P, editors. *Bioethics: An Anthology*. Oxford: Blackwell, 1999,243–54.

34 Wilkinson DJC, Savulescu J. Knowing when to stop: futility in the ICU. *Curr Opin Anesthesiol* 2011;24:160–5.

35 Schneiderman LJ, Jecker NS, Jonsen AR. Medical futility: its meaning and ethical implications. *Ann Intern Med* 1990;112:949–54.

36 Brody BA, Halevy A. Is futility a futile concept? *J Med Philos* 1995;20:123–44.

37 Helft PR, Siegler M, Lantos J. The rise and fall of the futility movement. *N Engl J Med* 2000;343:293–6.

38 Babnik J. Ethical decisions in the delivery room. *Textbook of Perinatal Medicine*. London: The Parthenon publishing group Ltd; 1998. 1880–5.

39 Chervenak FA, McCullough LB. The fetus as a patient: an essential ethical concept for maternal-fetal medicine. *J Matern Fetal Med* 1996;5:115–9.

40 Fenton LJ. Trisomy 13 and 18 and quality of life: treading softly. *Am J Med Genet A* 2011;155A:1527–8.

41 Yorgason B. *One Tattered Angel: A Touching True Story of the Power of Love*. Salt Lake City: Shadow Mountain, 2003.

42 Gabbay E, Calvo-Broce J, Meyer KB, Trikalinos TA, Cohen J, Kent DM. The empirical basis for determinations of medical futility. *J Gen Intern Med* 2010;25:1083–9.

43 Farlow B. Misgivings. *Hastings Cent Rep* 2009;39:19–21.

44 Koogler TK, Wilfond BS, Ross LF. Lethal language, lethal decisions. *Hastings Cent Rep* 2003;33:37–41.

45 McCaffrey MJ. Lethality begets lethality. *J Perinatol* 2011;31:630–1; author reply 1–2.

46 Caplan A. Neutrality is not morality: the ethics of genetic counselling. In: Bartels D, LeRoy B, Caplan A, editors. *Prescribing Our Future: Ethical Challenges in Genetic Counselling*. Hawthorne, NY: Aldine De Gruyter; 1993. pp. 149–65.

47 Elwyn G, Gray J, Clarke A. Shared decision making and non-directiveness in genetic counselling. *J Med Genet* 2000;37:135–8.

48 Resta RG. Eugenics and nondirectiveness in genetic counseling. *J Genet Couns* 1997;6:255–8.

49 Rentmeester CA. Value neutrality in genetic counseling: an unattained ideal. *Med Health Care Philos* 2001;4:47–51.

50 Williams C, Alderson P, Farsides B. Is nondirectiveness possible within the context of antenatal screening and testing?. *Soc Sci Med* 2002;Feb;54:339–47.

51 Bernhardt BA. Empirical evidence that genetic counseling is directive: where do we go from here? *Am J Hum Genet* 1997;60:17–20.

52 de Crespigny L. Words matter: nomenclature and communication in perinatal medicine. *Clin Perinatol* 2003;30:17–25.

53 McCormick RA. To save or let die. The dilemma of modern medicine. *JAMA* 1974;229:172–6.

54 Spinnato JA, Cook VD, Cook CR, Voss DH. Aggressive intrapartum management of lethal fetal anomalies: beyond fetal beneficence. *Obstet Gynecol* 1995;85:89–92.

55 Janvier A, Okah F, Farlow B, Lantos JD. An infant with trisomy 18 and a ventricular septal defect. *Pediatrics* 2011;127:754–9.

56 Leuthner SR. Palliative care of the infant with lethal anomalies. *Pediatr Clin North Am* 2004;51:747–59, xi.

57 Sanders RC. Prenatal ultrasonic detection of anomalies with a lethal or disastrous outcome. *Radiol Clin North Am* 1990;28:163–77.

58 Davidson N, Halliday J, Riley M, King J. Influence of prenatal diagnosis and pregnancy termination of fetuses with birth defects on the perinatal mortality rate in Victoria, Australia. *Paediatr Perinat Epidemiol* 2005;19:50–5.

59 Cunniff C, Kirby RS, Senner JW, Canino C, Brewster MA, Butler B, et al. Deaths associated with renal agenesis: a population-based study of birth prevalence, case ascertainment, and etiologic heterogeneity. *Teratology* 1994;50:200–4.

60 Parker SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE, et al. Updated National Birth Prevalence estimates for selected birth defects in the United States, 2004-2006. *Birth Defects Res A Clin Mol Teratol* 2010;88:1008–16.

61 Jaquier M, Klein A, Boltshauser E. Spontaneous pregnancy outcome after prenatal diagnosis of anencephaly. *BJOG* 2006;113: 951–3.

62 Kalucy M, Bower C, Stanley F, Burton P. Survival of infants with neural tube defects in Western Australia 1966-1990. *Paediatr Perinat Epidemiol* 1994;8:334–51.

63 Obeidi N, Russell N, Higgins JR, O'Donoghue K. The natural history of anencephaly. *Prenat Diagn* 2010;30:357–60.

64 Baird PA, Sadovnick AD. Survival in infants with anencephaly. *Clin Pediatr* 1984;23:268–71.

65 McAbee G, Sherman J, Canas J, Boxer H. Prolonged survival of two anencephalic infants. *Am J Perinatol* 1993;10:175–7.

66 Stoll C, Dott B, Roth MP, Alembik Y. Birth prevalence rates of skeletal dysplasias. *Clin Genet* 1989;35:88–92.

67 MacDonald IM, Hunter AG, MacLeod PM, MacMurray SB. Growth and development in thanatophoric dysplasia. *Am J Med Genet A* 1989;33:508–12.

68 Baker KM, Olson DS, Harding CO, Pauli RM. Long-term survival in typical thanatophoric dysplasia type 1. *Am J Med Genet A* 1997;70:427–36.

69 Vendola C, Canfield M, Daiger SP, Gambello M, Hashmi SS, King T, et al. Survival of Texas infants born with trisomies 21, 18, and 13. *Am J Med Genet A* 2010;152A:360–6.

70 Lebel RR, Roberson J, Van Dyke DL. Regarding trisomy 18. *Am J Med Genet A* 2006;140:964–5.

71 Bhanumathi B, Goyel N, Mishra Z. Trisomy 18 in a 50-year-old female. *Indian J Hum Genet* 2006;12:146.

72 Redheendran R, Neu RL, Bannerman RM. Long survival in trisomy-13-syndrome: 21 cases including prolonged survival in two patients 11 and 19 years old. *Am J Med Genet A* 1981;8:167–72.

73 Tunca Y, Kadandale J, Pivnick E. Long-term survival in Patau syndrome. *Clin Dysmorphol* 2001;10:149.

74 Szabó N, Gergev G, Kóbor J, Szucs P, Túri S, Sztriha L. Holoprosencephaly in Hungary: birth prevalence and clinical spectrum. *J Child Neurol* 2011;26:1029–32.

75 Veneselli E, Biancheri R, Di Rocco M, Fondelli MP, Perrone MV, Donati PT. Unusually prolonged survival and childhood-onset epilepsy in a case of alobar holoprosencephaly. *Childs Nerv Syst* 1999;15:274–7.

76 Stashinko EE, Clegg NJ, Kammann HA, Sweet VT, Delgado MR, Hahn JS, et al. A retrospective survey of perinatal risk factors of 104 living children with holoprosencephaly. *Am J Med Genet A* 2004;128A:114–9.

77 Yilmaz Z, Sahin F, Tarim E, Kuscu E. Triploidies in First and Second Trimesters of Pregnancies in Turkey. *Balkan J Med Genet* 2007;10:71–6.

78 Takabachi N, Nishimaki S, Omae M, Okuda M, Fujita S, Ishida F, et al. Long-term survival in a 69, XXX triploid premature infant. *Am J Med Genet A* 2008;146A:1618–21.

79 Bianchi DW, Bianchi DWF. *Fetology : Diagnosis and Management of the Fetal Patient*, 2nd edn. New York: McGraw-Hill Medical, London : McGraw-Hill [distributor], 2010.

80 Bae J, Jang M, Park S. Prolonged survival to adulthood of an individual with hydranencephaly. *Clin Neurol Neurosurg* 2008;110: 307–9.

81 Merker B. Life expectancy in hydranencephaly. *Clin Neurol Neurosurg* 2008;110:213–4.

Commentary on 'Professional responsibility in the perinatal management of ''lethal congenital anomalies'''

Drs Wilkinson, Thiele, Watkins and De Crepigny's paper on the concept of 'lethal congenital anomalies' calls to mind Socrates' complaint in *Euthyphro* that vague, imprecise concepts are like the statues of Daedalus: they do not 'stand fast and hold their ground' but wander about, resulting in confused and inapplicable judgements (Plato *Euthyphro* 11e). The concept of 'lethality', Wilkinson et al. correctly show, 'obscures rather than aids communication and counselling' of pregnant women about the management of pregnancies complicated by lethal fetal anomalies. To prevent this outcome—and the preventable confusion that it causes for pregnant women and among members of the health-care team—Wilkinson et al. propose to replace the discourse of 'lethality' with 'five key questions that are the basis for understanding the fetus's prognosis.' Wilkinson et al. identify four different meanings of 'lethality' in the current literature of perinatal medicine and ethics.

In our view, there is common to all four meanings a single concept: a lethal condition, properly understood, invariably leads to death, i.e. there is no effective treatment that will prevent a condition, disease or injury from causing death, in the near future. Failure to recognise that the 'near future' can become transformed by life-sustaining treatment in the neonatal intensive-care unit into an indefinite future is an outcome that the obstetrician should prevent. When there is a very high probability or near certainty of diagnosis of a fetal anomaly that results in death despite aggressive obstetric management and neonatal intensive care, the obstetrician, in conjunction with the neonatologist, should offer nonaggressive obstetric management followed by nonaggressive neonatal management (Chervenak et al., *Br J Obstet Gynaecol* 1995;102:434–5; 1999;106:293–6).

In our view, the question that Wilkinson et al. ask about physical and cognitive impairment can be more precisely formulated. When there is a very high probability or near certainty of a diagnosis that results in severe and irreversible loss of cognitive developmental capacity, the obstetrician and neonatologist should offer nonaggressive management (Chervenak et al., *Br J Obstet Gynaecol* 1995;102:434–5; 1999;106:293–6).

Their question about the burdens of treatment applies especially after the trial of neonatal critical care has been initiated and appears not to be succeeding. In our view, this question can also be more precisely stated. When the burdens of treatment overwhelm what little physical or cognitive developmental capacity the newborn may have, i.e. when the infant is only struggling to survive (McCormick *JAMA* 1974;229:172–6), then the neonatologist should recommend that the trial of intervention should be discontinued.

In summary, Wilkinson et al. should be congratulated for addressing a very important and timely topic. The ethical challenges created by a diagnosis of a lethal fetal anomaly are responsibly managed when obstetricians and neonatologists assert their professional responsibility to offer or recommend nonaggressive management (Chervenak et al., *Am J Obstet Gynecol* 2011;205:315.e1–5). ■

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