Evolving medical and surgical management of infants with trisomy 18

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Purpose of review
To review the evolving management of infants/children with trisomy 18, the prognosis with and without medical intervention, the factors that have contributed to the evolving management strategies, and an approach to the formulation of healthcare management plans for newborns with trisomy 18.

Recent findings
There has been a trend from nonintervention for infants/children with trisomy 18 toward management to prolong life. It has become clear that the prognosis for infants/children with trisomy 18 is not as 'hopeless' as was once asserted. However, case series of patients with trisomy 18 managed with a goal of prolonging life are not adequate to evaluate the efficacy of these interventions. They are also not adequate to support the contention that they have no efficacy. In fact, anecdotal evidence and medical plausibility suggest that treatment can prolong life in some cases. This trend has been supported by a change in emphasis from a largely physician-directed model of medical decision-making to a collaborative model, which respects parents' rights to make healthcare decisions for their children and recognizes that judgments about outcomes are often subjective, and social networks, which support and advocate for children with trisomy 18 and their families. An approach to collaborative medical decision-making that is goal-directed is recommended.

Summary
Healthcare management approaches or policies that reject out of hand the goal of prolonging the life of any infant/child with trisomy 18 are not defensible. Management plans should be goal-directed, based on the physician–parent evaluation of the benefits and burdens of care options for the individual child.

Keywords
general surgery, intensive care, thoracic surgery, trisomy 18

INTRODUCTION

In this chapter, we review how the management of infants/children with trisomy 18 has evolved, the prognosis for trisomy 18 with and without medical intervention, and the factors that have contributed to evolving management strategies, and finally we provide an approach to the formulation of healthcare management plans for newborns with trisomy 18.[1]

CHANGE IN THE APPROACHES TO MANAGEMENT OF NEWBORNS WITH TRISOMY 18

Beginning in the 1990s, a shift occurred from nonintervention toward management to prolong life. This shift is reflected in *Smith's Recognizable Patterns of Human Malformations*. In the fourth edition (1988) [1], Jones recommended 'limitation of all medical means for prolongation of life.' In the fifth edition (1997) [2], he recommended 'limitation of extraordinary means for prolongation of life should be seriously considered. However, the personal feelings of parents and the individual circumstance of each infant must be taken into consideration'. Recent professional surveys indicate a general movement on the part of neonatologists toward the latter recommendation [3,4].

Some sense of the interventions employed in the care of children with trisomy 18 in the United States and Canada can be gleaned from the Support

[1] The focus of this article is on infants/children with full trisomy 18. Trisomy mosaicism and partial trisomy are associated with quite variable outcomes – from near normal to the full trisomy 18 phenotype.

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KEY POINTS

- Life-prolonging medical treatment is increasingly being provided to infants with trisomy 18; there is evidence that neonatologists' attitudes are shifting toward a willingness to offer life-prolonging treatment.
- Even in an era of increased intervention, 1-year mortality with trisomy 18 remains very high – at least 75% even with aggressive and burdensome treatments, including surgery for CHD.
- Survivors are severely developmentally delayed, but routinely achieve cognitive and motor milestones; they are described as happy by parents and commonly enrich the lives of their families.
- Parents have become empowered by the Internet, support groups, and the disability rights movement to exercise parental autonomy in medical decision-making for trisomy 18 patients.
- Management plans should be goal-directed, collaborative, and transparent; should incorporate the values of the family as well as the medical facts; and strategically defer burdensome treatments until it is likely that patients will survive early infancy.

Organization for Trisomy 18, 13, and Related Disorders (SOFT) Surgery Registry [5]. One hundred and thirty-two surgeries on children with trisomy 18 for congenital heart defects (CHD) are documented. Most were atrial septal defect (ASD) (19) or ventricular septal defect (VSD) (33) repairs or patent ductus arteriosus (PDA) ligations (29). No surgeries are registered for repair of transposition of the great arteries, truncus arteriosus, or total anomalous venous return or specific documentation of surgical palliation for hypoplastic left heart syndrome. Five hundred and thirty-five other surgeries are documented in this Registry, most commonly gastrostomy (149), fundoplication (36), spinal fusion (23), cleft lip/palate repair (22), tracheostomy (19), placement of pressure equalization tubes (14), esophageal atresia/fistula (EA/TEF) repair (13), repair of strabismus (11), and inguinal hernia repair (10). Two surgeries for diaphragmatic hernia are registered, but none for omphalocele, gastroschisis, anal atresia, neural tube defect, nephroblastoma, or Wilm's tumor.

'NATURAL' HISTORY/PROGNOSIS

Survival

Eight population-based studies, spanning live births from 1968 to 2007, have reported survival rates of children with cytogenetically confirmed full trisomy 18 (Table 1) [6–13]. When discussing survival, the term 'natural' must be used advisedly because there are no data about the types of support provided in the neonatal period (see Bruns [14] for examples of varied support), although most were from eras in which surgical intervention was rare.

Seven of eight studies reported median survivals of 2.5–7 days; one [11] reported a median survival of 14.5 days. Overall, average survival was 42% at 1 week and 29, 12, and 8% at 1, 3, and 6 months, respectively. One-year survival ranged from 0 to 12%; the average was 4%. Only one study reported survival beyond 1 year – survival to age 6 years was 3% [9]. There were no significant changes in survival over time. Females consistently survived longer than males.

Even when reported, the causes of mortality in these population-based studies are too nonspecific to be informative. All report no relation between death and CHD. However, this is hardly surprising – most infants do not survive beyond 6 months and the most common types of CHD (VSD, ASD, PDA) associated with trisomy 18 [15,16▪] are unlikely to cause death in the first 6–12 months of life.

Prolonged survival

There are multiple reports in the English literature of survival beyond 5 years with trisomy 18, most to 12–20 years of age, but as long as 33 years [9,14,17–27,28▪▪]. Most of the larger case series recruited families/children from trisomy 18 support websites [14,20,24,28▪▪]. Without the number of live births with trisomy 18 during the time interval and the likelihood of selection/reporting bias, these reports only document that prolonged survival is possible, not its likelihood. In the population-based sample of Root and Carey [9], however, two of the three children who survived to 1 year were still alive at age 6. Thus, a substantial proportion of those alive at 1 year may survive beyond 5 years.

Neurodevelopmental outcome

Descriptions of long-term survivors in these case series confirm that they have severe to profound neurodevelopmental delay, but do slowly attain some milestones over time. None achieve expressive language and most are not capable of walking.

Baty *et al.* [29] described the psychomotor development of 62 children with trisomy 18, age 1–19 years. The ages at which milestones are achieved compared with the normal are presented in Table 2 [29]. Braddock *et al.* [28▪▪] assessed the ability of eight children with trisomy 18, age 4–29 years, to communicate. Receptive language skills were better than expressive communication

Table 1. Population-based survival rates of children with full trisomy 18

First author Source population	Carter *et al.* [6] Queensland, Australia	Young *et al.* [7] Leicestershire, UK	Goldstein and Nielsen [8] Denmark	Root and Carey [9] Utah, USA	Brewer *et al.* [10] Glasgow/ Scotland	Rasmussen *et al.* [11] Atlanta, USA	Vendola *et al.* [12] Texas, USA	Irving *et al.* [13] Northern Health Region, UK	
Years of births	1972–1982	1980–1985	1977–1986	1979–1988	1974–89/ 1989–97	1968–1999	1999–2003	1985–2007	
Live births	43	21	76	64	84	114	200	301	Total survivors/LBs
Survival									
1 week	15 (35%)	7 (32%)	33 (44%)	29 (45%)	36 (43%)	57 (50%)	104 (52%)	108 (36%)	389/903 (42%)
1 month	5 (11%)	4 (18%)	24 (32%)	22 (34%)	21 (25%)	43 (38%)	60 (30%)	81 (27%)	264/903 (29%)
3 months	2 (5%)	2 (10%)	4 (9%)	13 (20%)	ns	19 (17%)	ns	ns	40/318 (12%)
6 months	2 (5%)	0 (0%)	2 (3%)[a]	6 (9%)	ns	14 (12%)	ns	ns	24/318 (8%)
1 year	0 (0%)	0 (0%)	ns	3 (5%)	2 (2%)	10 (8%)	6 (3%)	18 (6%)	39/827 (4%)
6 years	ns	ns	ns	2 (3%)	ns	ns	ns	ns	
Median (days)	5	2.5	6	4	6	14.5	7	ns	

LBs, live births; ns, not specified.
[a]survivors at 5 months.

skills. None of the children were observed to produce any word approximations, but all exhibited augmentative and alternative communication behaviors.

Family experience

Some information has begun to emerge in the medical literature about the experiences of children with trisomy 18 or 13 and their parents [30▪▪]. These authors surveyed parents of 216 children with full trisomy 18 or 13 recruited from social support networks. Eight percent of these parents reported that they would not continue the pregnancy if another child was diagnosed with trisomy 18 or 13 prenatally; 9% reported that they would; and 83% reported that they would or would not have prenatal testing. Sixty-eight percent felt that the child had a positive effect on their marriage; 3% reported that their marriage had ended. Eighty-two percent of 160 parents with older children felt that the child had a positive effect on siblings. Forty-four percent of parents whose infant/child survived beyond 1 year reported that financial sacrifices were challenging.

Of the 187 parents whose children died, 88% felt that their child's overall experience was positive. Of the 159 parents whose children died after 3 months, 50% felt that their child had experienced more pain than other children and 50% felt that caring for their child was more difficult than expected; nevertheless, 98% reported that their child enriched their lives.

One hundred and four newborns received comfort care; one-third died before discharge and one-third lived beyond 1 year. Eighty-eight percent of parents of these children described the child's overall experience as positive, 68% had no regrets, and 31% regretted not considering more intervention. Of 53 newborns who received full support, one-third died before discharge and half survived beyond a year. Eighty-nine percent of parents of these children described their child's overall experience as positive; 89% felt that they had made the 'right' decision with no regrets. Ninety-eight percent of 79 living children (median age 4 years) lived at home. Ninety-nine percent of these parents of these children described their child as 'happy'.

PROGNOSIS WITH 'INTENSIVE' MANAGEMENT

In 1990, in a selected case series of six children with trisomy 18 who survived beyond 1 year, Van Dyke and Allen [20] reported the first two cases of surgery for CHD. One child underwent repair of a VSD and supravalvular pulmonary stenosis for congestive heart failure (CHF) at 29 months of age, but died in the postoperative period. The second child had a

Table 2. Developmental milestone achievements by 62 children with trisomy 18

Milestone	Age achieved[a] (months)	Age range (months)	Number achieving milestone	Normal age range achieved
Smiled responsively	4.7 (0.5)	0.5–24	54	0–2
Held head up	9.0 (1.5)	0.3–36	33	0–2.5
Watched toy or face	4.4 (0.6)	0.2–24	57	0–1
Reached for toy	9.6 (1.2)	2.5–36	38	3–5
Laughed out loud/giggled	13.0 (3.1)	2.3–96	36	1.5–3.3
Sat up with help	20.4 (2.9)	3.5–60	25	1.6–4.3
Sat up with alone	38.5 (6.3)	7.5–72	12	4.8–7.8
Said consonant sounds	23.0 (6.2)	8.0–52	8	5.6–10
Rolled over	30.5 (16.5)	0.2–540	32	2.2–4.7
Balanced on hand and knees	53.7 (18.1)	12–204	10	–
Walked in walker	39.5 (7.4)	24–60	5	–
Cruised furniture	72	–	1	7.4–12.7
Walked unaided	–	–	0	11.2–14.4
Used signs	61.5 (9.9)	36–84	4	–
Number of words	3.4 (0.7)[b]	1–5 years	5	–

Modified with permission from [29].
[a]months-mean (standard error).
[b]number of words.

VSD and PDA closure at 2 years of age; she was alive in a special school program at the time of last follow-up at age 7.5 years. Four other children had VSDs, which were not repaired. One died of unrelated causes, another with severe pulmonary hypertension died suddenly at 23 months of age, and two died with severe CHF in the second year of life. Although anecdotal, this case series suggested more than 20 years ago that two prevailing beliefs – that surgery for CHD would not prolong the life of children with trisomy 18 and that children with trisomy 18 with CHD do not die as the result of their CHD – were mistaken.

There are two reports of outcomes of newborns with trisomy 18 who received supportive care to prolong life [31,32]. In the first [31], 20 cases with full trisomy 18, cared for in one neonatal intensive care unit (NICU) in Poland, received 'full care and diagnostic procedures' for 2 weeks until the diagnosis was confirmed cytogenetically. After confirmation, therapies were restricted to palliative surgery for CHD and 'aggressive' therapy for respiratory failure, circulatory failure, and severe infections.

Nineteen infants had CHD. Three with ductal dependent lesion received prostaglandin E until the diagnosis was confirmed and four had surgery to limit pulmonary blood flow. None had surgery for ductal dependent CHD. Sixty-five percent had central nervous system malformations; only one required surgery (for a neural tube defect) in the immediate newborn period. Management of hydrocephalus after confirmation of the diagnosis was not specified. Of five patients with other major congenital anomalies, two had diaphragmatic hernia repairs, one omphalocele repair, and one EA repair. Four of six survivors and five of nine nonsurvivors without ductal dependent CHD underwent surgical procedures.

Median time to death was 20 days. Six survived beyond 1 month. The authors concluded that 'despite aggressive treatment most ... died in the neonatal period'.

Kosho *et al.* [32] reported outcomes of 24 newborns with trisomy 18 (without sufficient testing to exclude mosaicism) cared for in one NICU in Japan, who were 'managed under the principle of providing intensive treatment' with informed parental permission. This included delivery room resuscitation, mechanical ventilation, blood products, parenteral nutrition, corrective or palliative gastrointestinal surgery to establish enteral nutrition, and medical therapy for CHF.

Median survival was 152.5 days. Ninety-six percent of newborns survived one day, 88% 1 week, 83% 1 month, and 25% for at least 1 year. The longest survivor died at 58 months because of tracheostomy tube complication. At the time of publication, there was only one survivor. The authors concluded that these results 'suggested improved survival compared with previous population-based studies' but acknowledged the possibility of selection bias.

Since 2004, four case series of a total of 90 children who underwent surgery for CHD have been published in the English literature (Table 3) [33–36]. All but 24 patients are from Japan. Three of the case series did not specify whether cases with trisomy mosaicism were excluded; in the fourth, three of nine cases had trisomy mosaicism. By far, the most common surgery was pulmonary artery banding, frequently with concomitant PDA ligation. There were a few cases of primary VSD closure, a few cases of closure of VSD subsequent to pulmonary artery banding, and a few cases of repair of coarctation of the aorta. There were no cases of repair of tetralogy of Fallot (TOF) in the three studies from Japan. There were no reported cases of surgery for more complex CHD. The wide variation in age at surgery and lack of controls precludes evaluating the efficacy of surgery for CHD.[2] However, the lengths of stay suggest that postoperative courses were more complicated than expected and hospital survival was lower than expected for the surgeries performed compared with otherwise normal children.

FACTORS CONTRIBUTING TO EVOLVING MANAGEMENT STRATEGIES

Particularly relevant to arguments for or against life-prolonging treatment of these children are the shift in decision-making authority from physician to family; improvements in the medical management of children with complex medical needs; and disability advocacy, the proliferation of support groups for patients with rare illnesses, and the rise of the Internet.

Parental authority

The ascent of the principle of respect for patient autonomy has been mirrored in the United States by a shift to strong parental authority in healthcare decisions for their children. This shift is founded on the expectation that parents know their children better than anyone and are motivated by close emotional ties to act in the child's best interest. The limits of parental autonomy are poorly defined, but are typically relevant only when parents make decisions 'clearly' against a child's best interests, causing substantial harm, suffering, or death – and this determination can withstand the scrutiny of judge and jury.

Essential to the exercise of parental authority is the disclosure of all information necessary to make sound decisions. The standard for disclosure has shifted away from a professional standard, whereby the information customarily disclosed by physicians must be presented, to a reasonable person standard, according to which the information that a hypothetical, reasonable (and nonmedical) person would find pertinent to the decision must be disclosed.

Improvements in medical treatment

Major improvements in neonatal and pediatric critical care have resulted in substantial reductions in mortality over the past 20 years. Over the same time period, the number of children with complex and chronic conditions treated in ICUs has increased substantially. The availability and efficacy of treatments used to prolong the life of chronically ill patients have had an inevitable impact on the medical and ethical arguments used to defend management strategies for trisomy 18 patients.

Disability advocacy, patient support groups, and the Internet

The impact of modern communications, particularly the Internet, on the experience, support, and treatment of patients with rare diseases and their families cannot be overstated. Whereas 20 years ago a family facing the birth of a baby with trisomy 18 would be largely dependent on the knowledge and expertise of the immediately available healthcare providers, today most families have access to a wealth of information as well as access to support group websites, such as SOFT [37]. These groups are also commonly willing to advocate for the rights of these patients and their families.

Ethical justifications for nontreatment

Arguments against life-prolonging treatment of these babies focus on the harms caused by these interventions. The obligation to do no harm, as well as the moral distress and threat to professional integrity in providing burdensome treatment with limited benefit, shape the justification for not offering life-prolonging treatment.

These justifications have been criticized for several reasons. It has been pointed out that the best interests calculation contains concealed quality of life judgments that may devalue the lives of patients with trisomy 18 [38]. Additionally, critics note that the lack of benefit used to justify nontreatment is historically based, and that there is a self-fulfilling prophecy evident when high mortality is assured by not offering life-prolonging treatment [39].

[2] Although Maeda *et al.* [36] compare survival of children with trisomy 18 and CHD who underwent surgery with those who did not during the same time period, selection bias (due to indication for surgery and parent refusal of surgery) precludes interpretation of the difference.

Table 3. Case series of infants/children with trisomy 18 who underwent surgery for congenital heart defects[a]

	Graham *et al.* [33]	Kaneko *et al.* [34]	Muneuchi *et al.* [35]	Maeda[b] *et al.* [36]
N	**24**	**17**	**9**	**32**
Limited to full trisomy 18	ns	ns	3/9 with mosaicism	ns
Age at surgery	145 days (6 days–6 years)	66 days (7–258 days)	50 days (14–195 days)	ns
CHD				
VSD	14[c]	3	1	15
ASD	0	0	0	0
PDA	2[c]	0	1	0
VSD with ASD and/or PDA	ns[c]	9	5	10
AVSD	1	0	0	0
TOF	5	0	0	1
AoCoarct	2[c]	0	0	0
VSD and PDA with AoCoarct	0	4	1	4
ASVD and PDS with AoCoarct	0	1	0	0
DORV	0	0	1[d]	2[e]
Surgery				
VSD closure	ns[f]	0	0	3
PA banding	ns[f]	8	5	18
PA banding with later VSD closure	ns[f]	2	0	0
VSD closure and PDA ligation	ns[f]	2	0	1
VSD and/or ASD closure	ns[f]	0	3	0
PDA ligation	ns[f]	(14)[g]	1	(7)[h]
AoCoarct repair	ns[f]	0	0	0
PA banding and AoCoarct repair	ns[f]	4[i]	0	3
VSD and AoCoarct repair	ns[f]	1	0	1
Systemic to PA shunt	ns[f]	0	0	2
TOF repair	ns[f]	0	0	0
Length of stay (days)	9 (4–27)	ns	142 (20>1996)	ns
Survived hospitalization	21	14	5[j]	ns
Survivors at time of data collection (age range of survivors at time of data collection)	No post discharge follow-up	6 (99–1384 days)	6 (20–1996 days)	18 (ns)

AoCoarct, aortic coarctation; ASD, atrial septal defect; AVSD, atrial ventricular septal defect; d, days; DORV, double outlet right ventricle; ns, not specified; PA, pulmonary artery; TOF, tetrology of Fallot.

[a]The four cases of trisomy 18 who underwent surgery for CHD in Kaneko *et al.* [43] were not included in the table because these cases appear to be included in Kaneko *et al.* [34]. The 24 cases of trisomy 18 who underwent surgery for CHD in Yamagishi [44] were not included in the table because of the possibility that cases from Kaneko *et al.* [34], Muneuchi *et al.* [35] and Maeda *et al.* [36] may have been included.

[b]In this series, it was not known what surgery was performed in four cases.

[c]Some of these cases had additional cardiac defects that were not specified.

[d]In this case, the DORV was associated with a VSD and PDA.

[e]One case of DORV was associated with pulmonary stenosis.

[f]This report included cases with trisomy 13; data were not provided separately for cases with trisomy 18.

[g]Eleven PDA ligations were done concomitantly with PA banding (in two of these cases the VSD was subsequently closed) and three with VSD closures.

[h]Seven PDA ligations were done concomitantly with PA banding.

[i]Two of these cases subsequently underwent VSD repairs.

[j]One survivor remained hospitalized at 1996 days.

Ethical justifications for treatment

Those who favor offering life-prolonging treatment argue that it is the parents' right to determine what is in the best interest of their infant/child. Proponents assert that patients with trisomy 18 can survive infancy, enjoy a reasonable quality of life as judged by their families, and are loved members of the family.

Critics point out that acceding to parents' wishes for life-prolonging treatment in the majority

of cases of trisomy 18 will increase suffering with limited benefit and foster a consumer model of medicine where families are able to elect treatments regardless of the likelihood of success.

APPROACHES TO FORMULATING MANAGEMENT PLANS

Terms often used to describe the prognosis of infants with trisomy 18 – such as 'lethal, 'imminent death', 'terminal state', 'futile', and 'hopeless' – are value-laden and inaccurate. Terms used to describe management or interventions – such as 'intensive', 'aggressive', 'extraordinary', or 'appropriate' – are too vague to be useful in formulating management plans. Rather, this formulation should be goal-directed.

The first step in this process is to define the goal(s) of management. Goals may span a spectrum from maximizing comfort and minimizing burden without attempting to prolong survival, to facilitating discharge home without otherwise prolonging survival, to prolonging survival to the greatest extent possible. Goal(s) should be developed jointly by informed parents and the physician. The duty of parents in developing goals of care is to give due consideration to the best interests of their child. The duty of the physician is to facilitate the proper exercise of parental authority, but also to fulfill the fiduciary duty to protect the child from harm and suffering that is not in their best interests.

The next step is to evaluate the likelihood that the goal is achievable. This is largely a medical judgment. However, there are limited data to inform this evaluation because survival is relatively limited regardless of management and the efficacy of interventions to facilitate discharge home or prolong survival is largely unproven. However, some goals – such as allowing the child to eventually live independently should he or she survive – are clearly not achievable. It would be helpful to predict who might benefit most from interventions. Those who will survive beyond 1 year would be the most likely to benefit from management to prolong and maximize the quality of life. However, the small number of survivors beyond 6–12 months precludes the identification of any characteristic(s) that might be associated with prolonged survival to an extent that would be useful in informing management decisions [40], although female sex is consistently associated with longer survival [6,9,11,25,41]. Given this inability to predict survival, consideration should be given to deferring burdensome intervention (unless necessary for comfort or to prevent death) until it becomes more likely that the child will survive beyond 6 months or 1 year. For example, in the absence of EA/TEF, gastrostomy tube placement could be deferred for weeks or months. On the other hand survival beyond the newborn period with EA/TEF is unlikely if parenteral nutrition or some intervention to allow enteral feeding (e.g. gastrostomy) is not provided. CHD does not predict early mortality. The most common types of CHDs in children with trisomy 18 are not likely to cause 'early mortality' and can be managed medically for some period of time. Therefore, pulmonary artery banding or VSD/ASD/PDA closure could be deferred until 6 months to 1 year.

Finally, a value judgment must be made about whether the goal, even if achievable, is 'worth it' in terms of the benefits to the child and in terms of the just distribution of limited societal resources. In neither case is the decision primarily a medical judgment. At the individual patient's level, respect for parental autonomy warrants serious consideration of the informed family's judgment about what is in the best interests of their infant. Judgments about the equitable distribution of limited resources should be made at the societal level as policy decisions in order to prevent injustices in the treatment of individual patients [42].

CONCLUSION/RECOMMENDATIONS

Management strategies that deny the possibility that some children with trisomy 18 can benefit from life-prolonging treatment should be rejected. Likewise, strategies that deny the possibility that some children with trisomy 18 will be overly burdened by parental decisions to pursue life-sustaining treatment should be rejected. Effective strategies will begin with a shared decision-making model featuring bidirectional communication of medical facts and family values. Treatment decisions must then be based on realistic goals of care, individualized and based on the burdens and potential benefits of each treatment option for a particular baby. Importantly, prudent decision-making will often involve deferring burdensome treatments until it becomes more likely that the child will survive infancy. Finally, hospital policies addressing requests for inappropriate medical treatment of children may empower healthcare providers to appropriately advocate for their patients and to combat a 'consumer model' of healthcare.

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Conflicts of interest

There are no conflicts of interest.

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- ■ of special interest
- ■■ of outstanding interest

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