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**ABBREVIATION**

AVSD—atrioventricular septal defect

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## Should We Mend Their Broken Hearts? The History of Cardiac Repairs in Children With Down Syndrome

In May 2014, Evans et al<sup>1</sup> reported that children with Down syndrome had lower rates of in-hospital death after cardiac surgery, compared with children without Down syndrome. Forty years ago, these results would have been unthinkable, as heart defects were not repaired in the majority of children with Down syndrome. However, as the field of cardiac surgery evolved, equal postoperative outcomes were reported between children with Down syndrome and those without. The historical question of whether we ought to offer cardiac repairs to infants with Down syndrome was influenced by a complex web of ethical, social, and legal considerations that changed over time, resulting in the current standard of care in which children with and without Down syndrome have the same opportunity for cardiac repair.

Complex intracardiac surgery in infants was not routinely performed until the early 1970s, and *Pediatrics* published the first report on cardiac repairs in infants with Down syndrome in 1976.<sup>2</sup> The authors concluded that overall

surgical mortality in Down syndrome was high, especially in atrioventricular septal defects (AVSD), the most common heart defect associated with Down syndrome. Critical appraisal reveals only a minority of the children in the cohort received cardiac repairs, and of those, many were not suitable for full repairs because they had developed pulmonary hypertension by the time of surgery. In a 1978 Letter to the Editor,<sup>3</sup> Feingold suggested that children with Down syndrome did poorly because they did not receive prompt surgical repairs, and not because of their Down syndrome.

Over the following years, cardiac surgical techniques in infants developed rapidly, with improved outcomes in all children. The advent of echocardiography in the late 1980s allowed for screening and early diagnosis of cardiac defects in community settings, leading to earlier diagnoses and repairs.<sup>4</sup> Frid et al<sup>5</sup> traced rates of isolated AVSD repairs in children in Sweden with Down syndrome, finding increasing rates of repairs from 1973 to 1997, as well as improving short and

long-term mortalities (Table 1). The age of cardiac defect diagnosis in infants with Down syndrome also dramatically decreased, from 861 days of age in 1970 to 4 days in 1996.

Although the medical literature showed no increase in postoperative mortality in children with Down syndrome versus those without,<sup>6,7</sup> some of these children were denied surgical repairs into the 1990s. In 1985, Bull et al<sup>8</sup> argued that withholding cardiac surgery in a child with Down syndrome was a valid clinical decision. Given operative mortalities of ~20% at that time, the authors suggested that the early risk was justified in a chromosomally normal child but not in a child with Down syndrome, whose life expectancy and mental capacity were already limited. At that time, the majority of persons with Down syndrome could be expected to live into their 40s and 50s, whereas those with an unrepaired heart defect had survival rates of only 2% by 20 years of age.<sup>9</sup> The same year, Sondheimer et al<sup>10</sup> published a case series demonstrating that children with

**TABLE 1** Surgical Results of Isolated AVSD Repairs in Children With Down Syndrome in Sweden (Adapted from Frid et al)<sup>5</sup>

Year of Birth	Cardiac Repairs (%)	30-Day Mortality <sup>a</sup> (%)	5-Year Mortality <sup>a</sup> (%)
1973–1977	39.0	28.1	35.5
1978–1982	71.2	21.3	32.6
1983–1987	75.3	17.2	31.3
1988–1992	83.5	12.8	26.7
1993–1997	97.0	1.0	10.1

<sup>a</sup> There were no significant differences in early and late postoperative mortalities between Down syndrome and non-Down syndrome groups.

Down syndrome were denied access to cardiac surgery by way of delayed referral. All chromosomally normal children were referred and underwent repairs before 1 year of age, but 10 of 28 children with Down syndrome were referred after 1 year of age, with 5 being inoperable due to pulmonary hypertension.

Why did it take so long for cardiac repairs to become standard of care in children with Down syndrome? The social history of subjects with Down syndrome reveals an evolution of social factors that played as large a role in the provision of cardiac surgery as advancing surgical techniques (Figure 1). Eugenics movements in the 1920s led to the institutionalization and sterilization of persons with mental retardation, Down syndrome being the most common genetic cause.<sup>11</sup> Grassroots parents' associations formed in the 1950s and eventually coalesced into national Down syndrome societies, advocating for social supports, education, and research. In 1968, the first Special Olympics resulted in positive publicity for people with Down syndrome, celebrating their abilities; 1968 was also the year that amniocentesis techniques provided prenatal diagnoses

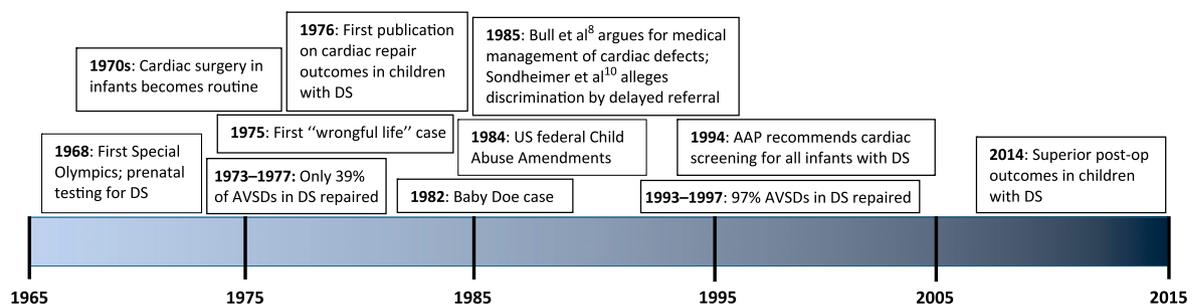
and the option for selective termination of fetuses with Down syndrome.<sup>12</sup>

The year 1975 brought the first “wrongful life” case, when a woman from New York successfully sued her physicians for not advising her of prenatal testing techniques that would have allowed her to terminate her pregnancy, which resulted in the birth of an infant girl with Down syndrome. In 1980, Baby Pearson died shortly after birth, rejected by his parents because of Down syndrome. The pediatrician, Dr Leonard Arthur, was acquitted of attempted murder charges, after ordering regular doses of dihydrocodeine and writing in the chart “Nursing care only. Parents do not wish it to survive.” Baby Doe, another newborn with Down syndrome, died after his parents were allowed to refuse life-saving gastrointestinal surgery by the Indiana courts in 1982, despite offers by 10 other couples to adopt the child. These cases illustrate society's willingness to accept the passive euthanasia of children considered to be “irreversibly disabled.”<sup>13</sup>

Similar attitudes played a role in the failure to offer cardiac repairs to children

with Down syndrome. In 1975, Crane<sup>14</sup> published surveys of pediatric cardiac surgeons who stated that they were more likely to repair a cardiac defect in a child with a cardiac defect and another physical anomaly versus a child with a cardiac defect and Down syndrome (90% vs 59% would usually operate). She also found that neither the severity of the cardiac defect, nor the impairments related to Down syndrome, affected the rates of repairs. Instead, having no siblings, being the product of a precious pregnancy, or having a family that requested surgery were positive predictors of receiving a repair. From the 1980s onward, public opinion gradually became more accepting of persons with Down syndrome. Both de-institutionalization and improved developmental and educational supports demonstrated the full potential of these individuals and allowed them more visibility in public life. The federal Child Abuse Amendments of 1984 in the United States made it illegal to withhold indicated medical treatment from children with disabilities.<sup>15</sup> The American Academy of Pediatrics' first health supervision guidelines for children with Down syndrome (published in 1994)<sup>16</sup> recommended routine cardiac screening of all infants with Down syndrome, suggesting that offering cardiac repairs to children with Down syndrome had become standard of care.

The lesson learned in the history of cardiac repairs in children with Down

**FIGURE 1**

Timeline of events leading to cardiac repairs in Down syndrome (DS). AAP, American Academy of Pediatrics.

syndrome is the degree to which social, ethical, and legal factors define the context of clinical decision-making. Once it was shown that children with Down syndrome had equal postoperative outcomes, it took almost 2 decades for cardiac repairs in children with Down syndrome to be fully accepted as standard of care.

The contemporary question of whether individuals with Down syndrome should be the recipients of heart transplants is the next chapter in whether we should advocate “mending their broken hearts.” Like our contemporaries in the 1970s, we acknowledge theoretical concerns related to Down syndrome (eg, increased risks surrounding posttransplant infections, lymphoproliferative disorders, pulmonary hypertension) and have little literature to guide us. Certainly, we should strive to avoid the historical pitfalls of discrimination against persons with Down syndrome, based on their intellectual disabilities or potential physiologic complications. This time, our emphasis should be on well-designed research and timely evidence-based guidelines to aid in such complex decision-making.

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