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Opinion

# On Objectivity in Prenatal Genetic Care

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**Abstract:**

We address an ongoing controversy over what health-care providers tell prospective parents about Down Syndrome (DS). Many parent, disability-rights, and anti-abortion activists believe that the messages that health-care professionals transmit to pregnant women and their partners are distorted. In their view, OB-GYNs, primary-care providers, clinical geneticists, midwives and other medical professionals generally assume that the quality of life for individuals with DS and their families is poor, whereas in fact, those with personal experience of DS are almost always satisfied with their lives. The critics believe that providers' biases, directly or indirectly communicated to prospective parents, explain high rates of pregnancy termination for DS. If the information were unbiased, the argument goes, these rates would fall. An underlying assumption of this argument is that information provided by those with experiential knowledge, who know what it is like to live with a particular condition, will be unbiased. However, we argue that there are grounds to be skeptical of this apparently plausible assumption. We also suggest strategies that we believe will advance the cause of achieving objectivity in prenatal care.

**Keywords**
Prenatal testing; Down syndrome; counseling; abortion; disability rights; disease advocacy organizations; “Dear Future Mom”; benefit-finding; adaptation

Many expectant mothers, informed after prenatal testing that their child will have Down Syndrome (DS), choose to end the pregnancy. Just how many make this choice is surprisingly hard to say. In some countries, such as the U.S., statistics are not collected on a national basis, and published studies may be demographically unrepresentative and also lag behind practice, overstating termination rates in places where rates have been falling. Thus, an oft-cited review, based on studies published between 1980 and 1998 [1], reported a termination rate of 92%, whereas a more recent meta-analysis of U.S. studies conducted between 1995 and 2011 [2] reported lower rates, which have likely continued to decline. Substantial national and regional differences as well as variability associated with race, ethnicity, maternal age, socio-economic status, and other variables also make it hazardous to generalize from existing analyses, based on a handful of studies, to entire countries or regions.

We do know that termination rates have generally declined, reflecting improvements in medical outcomes and life expectancy, cultural shifts toward greater acceptance and social integration of individuals with DS, the routinization of prenatal screening (and hence inclusion of more women disinclined to abortion), and in some countries, an increasing tendency to stigmatize abortion and also to erect legal and practical barriers to access to abortion services. Despite this trend, in many countries the practice of abortion for fetal anomaly, and especially DS, has become increasingly contentious.

There are multiple and perhaps not fully understood reasons for this development. A major factor is certainly the resurgent anti-abortion movement, especially in countries where right-wing, populist parties now dominate the government. In this context, bans on abortion for fetal anomaly represent partial successes where public opinion or the courts block outright prohibition. Thus, in Poland, the ruling Law and Justice party originally supported a ban on all abortion (currently legal only in cases of rape or incest, a serious threat to the mother’s health, or severe and irreversible damage to the fetus). Although it was ultimately retracted after mass protests, that bill was followed by another, currently before the Parliament, to ban abortion in cases of fetal anomaly [3]. Similarly, in the U.S., where activists’ focus has shifted from efforts to overturn Roe v. Wade to obtaining incremental restrictions on abortion, Prenatal Non-discrimination Acts (PRENDAs), which bar abortion based on determination of fetal sex, race, and most commonly, fetal anomaly, have become increasingly popular. Culturally, the representation of abortion as a dramatic act that can only be justified by extreme suffering of the child/mother/family (a phenomenon described in recent books by Katie Watson [4] and Carol Sanger [5]), has likely also contributed to discord over abortion sought to avoid fetal anomaly. When abortion is presented as defensible only in dire circumstances, its use to prevent the birth of a child with DS is almost bound to be viewed unfavorably.

Another factor is the increased visibility and influence of the disability-rights movement. Although movement activists do not necessarily oppose abortion *per se*, they tend to be uncomfortable with disability-selective abortion, which is often condemned as “eugenics” [6].

Disease-advocacy organizations, such as local, national, and international DS associations, have also mobilized politically and, like disability-rights groups, often adopt a critical stance towards selective abortion. Although anti-abortion, disability-rights, and disease-advocacy activists may differ in their general socio-political orientations, they are at times *de facto* allies on the issue of pregnancy termination based on determination of fetal anomaly [7, 8].

The fact that many women will choose to abort following a confirmed diagnosis of DS is often explained (or explained-away) as the result of misinformation transmitted by health-care providers. Thus, the British advocacy organization "Don't Screen Us Out" asserts that "The UK's obligations under the Convention on the Rights of Persons with Disabilities (CRPD), entail that disabled people and their families should be accommodated, included, and supported by society. Yet the evidence suggests that parents of children diagnosed with disability are not given the information and help they need to choose to bear and raise their disabled child" [9].

According to critics of disability-selective abortion, OB-GYNs, primary-care providers, clinical geneticists, midwives, and other health professionals generally believe that the quality of life for individuals with DS and their families is poor. Thus, various witnesses before the UK Parliamentary Inquiry into Abortion on the Grounds of Disability testified that changes "in the improved life expectancy, medical treatment and situation of Down's Syndrome children, and the achievements of individuals with learning difficulties, are not reflected in the attitudes of the medical profession towards parents and their child" [10, 11]. The critics maintain that medical professionals' assumptions are wrong, and that those with personal experience of the condition are in fact almost always satisfied with their lives. In the critics' view, providers' biases, directly or indirectly communicated to prospective parents, explain high rates of pregnancy termination for DS. If the information were unbiased, the argument goes, these rates would drastically fall. In the U.S., a "pro-information" movement based on these assumptions has attempted, with considerable success, to legislatively require that the information provided by health-care providers be objective [12, 13]. (We are here only concerned with critics' perceptions. The question of what providers *actually* think and the extent to which they act on their beliefs is immensely complicated since empirical studies indicate substantial variation both among types of medical professions—for example, the views of clinical geneticists and genetic counselors often diverge from those of primary-care providers—and among countries—for example, attitudes in the Netherlands differ starkly from those prevalent in France—and because many clinicians avoid discussion of prenatal issues altogether [14-17]).

Critics of current counseling practices often assume that information provided by those with experiential knowledge, who know what it is like to live with a particular condition, is truer to reality and should thus be privileged. But there are grounds to be skeptical of the assumption that patient/parent perspectives escape bias. An important reason is that, in practice, patient/parent perspectives are equated with the position of the organizations that claim to speak on their behalf. Yet organizational viewpoints may be unrepresentative of what is a wide range of individual experiences, perceptions, and viewpoints. Individuals with a hereditary condition may have quite diverse opinions about the transmission of the condition to their offspring. Thus, some people with hypohidrotic ectodermal dysplasia (HED), a hereditary condition linked to unusual facial features, absent or malformed teeth, and problems with sweating that can lead to dangerous fevers, wish to spare their children hardships that they experienced, while other believe that the presence of the same condition in their children would produce stronger familial bonds, and that a

relative's decision to select against HED reflects a disparaging judgment on the quality of their life [18]. However, a diversity of "experiential knowledges" about life with impairment may be easier to express in the absence of visible and active patients' associations that propagate—often through the voice of a small number of devoted and charismatic leaders—a strong opinion against all forms of "eugenic selection". Such associations, especially when they play an important role in promoting the rights of people with a given impairment, may persuade their members to adopt the view of the association's leadership, or, alternatively, make expression of dissenting opinions more difficult.

If providers exaggerate burdens, patient/parent advocacy groups tend to focus exclusively on infants and children, rather than adults, and on the most positive outcomes, ignoring the heterogeneity of patient and parent circumstances and attitudes and fact that DS, like many conditions, is characterized by a wide clinical spectrum. The older view of people with DS as invariably severely incapacitated has largely been replaced in public discourse with an upbeat but equally one-sided portrayal. The video "Dear Future Mom", developed for the 2014 World Down Syndrome Day by the Italian national DS advocacy organization CoorDown in collaboration with the advertising firm Satchi & Satchi Italy, is a case in point [19]. In this widely-watched video—which on its release broke records for social-media "shares" in a 24-hour period and has been viewed nearly eight million times since—15 people with Down Syndrome assure a pregnant woman who has just learned that her fetus is affected that her child will be able to speak, learn to read and write, attend school, hold a job, travel, rent an apartment and live alone. (Ironically, one of the few population-based studies of quality of life in DS was conducted in Italy and reports generally grim statistics on variables relating to adults, including low employment or involvement in any regular activity. The authors note that: "After the age of 30, the percentage of people demonstrating decline in function increased sharply, while disability-related support decreased" [20]).

Although a narrator acknowledges that "some days will be difficult", he immediately comments that this is true for all moms. There is no mention of cardiac and other medical complications, psychiatric issues, especially in high-functioning teenagers and adults, difficulties in controlling sexuality, issues related to older adults, such as early Alzheimers, the possibility that the child will never even learn to speak, or the problem of care after the parents are no longer able to provide it. The last is a particularly notable omission since worries about long-term care are a major motivation for those choosing to terminate a pregnancy after a diagnosis of DS [21], worries that can only intensify with increases in average life-expectancy (which in some countries has more than doubled in the last quarter-century, from a mean of 25 to about 60 years) [22]. In general, the question of care for those who are severely disabled or disturbed, and especially maternal care (since women provide most of the care-giving) is either unacknowledged or in effect dismissed with the observation that "society" should be obliged to care for all its disabled members [23].

Must we therefore conclude that the quest for objectivity is simply a will-o'-the-wisp, and that, wherever we look, we will inevitably encounter spin? While total objectivity may be unachievable, we believe that it is possible to more nearly approach that goal. One step in that direction, as the critics and several professional associations suggest, is for prospective parents to be told that affected individuals and their families generally rate their quality of life higher than do those who are asked to imagine what it would be if their child had the prenatally-diagnosed condition. That is

an important fact with potential relevance for reproductive decision-making. But how it should be interpreted is not self-evident.

The gap between insider and outsider ratings (or more technically, measured "experience utility" versus "hypothetical utility") is a general phenomenon, holding true for most chronic diseases and disabilities [24]. It is explained by two factors: the greater knowledge that insiders possess and also the process of adaptation that they typically undergo. In general, human beings are skilled at "benefit-finding"—perceiving silver linings and concluding that what initially seemed tragic was in retrospect a blessing (the experience having strengthened character, brought family and friends closer, and enhanced appreciation of the hitherto taken-for-granted). But it is also true that some parents do *not* adapt well, that personal and social circumstances as well as the nature of the condition matter, that adaption is a lengthy process that may entail much pain along the way, and not simply an end-point, and that the process may be positive and negative, or "constructive" and "resigned," to use terms coined by Peter Warr and Paul Jackson in their research on adaption to prolonged unemployment [25]. Thus, adaptation may involve learning new skills, participation in new activities, and the development of new interests and goals—but also lowered expectations, a passive acceptance of new circumstances, and cognitive denial.

Moreover, while the fact that most families ultimately adapt well to the birth of a child with a disability should be communicated to pregnant women and their partners, it is also important to acknowledge that this outcome is not universal, and that while some families will be strengthened by the experience, especially where the children typically have happy dispositions, others will not. The risk of family disruption in some disorders is significant. In a moving TEDx talk, Germana Soares, founder and president of the Uniao de Maes de Anjos (Union of Mothers of Angels) which includes about 400 mothers of children with congenital Zika syndrome (CZS), notes that 76% of the mothers in this association, including herself and the association's vice-president, were abandoned by their male partners after the birth of a CZS child [26].

Other elements of a way forward were already identified two decades ago by sociologists Aliza Kolker and Meredith Burke in their classic text on prenatal testing [27]. Kolker and Burke proposed that women be provided with examples of differing severity, with their relative frequency indicated. The point about relative frequency is crucial. Currently, even the most sophisticated pamphlets provided to pregnant women read like the inserts in drug packages that list numerous potential complications without any data on how often they occur.

The guidelines of the (U.S.) National Society of Genetic Counselors (NSGC) are illustrative. The NSCG has seriously wrestled with the issue of how to reconcile the differing perspectives of medical professionals and DS advocacy groups. Its publications laudably acknowledge the existence of challenges, and its best-practice guidelines recommend that prospective parents be offered an "opportunity to meet with families who are raising a child with Down syndrome, those who have chosen to create an adoption plan, and/or those who have terminated a pregnancy" [28]. (The last might be difficult to effect, especially in the U.S. Even in Britain, where the pro-life movement is less influential, it is increasingly difficult to openly acknowledge abortion for DS. Thus Jane Fisher, Director of the Antenatal Results and Choices organization, notes that for women who acknowledge terminating a DS pregnancy, "the vitriol and hate mail are mind-boggling" [29]). However, the NSGC guidelines include no quantitative data on outcomes in non-medical spheres such as education or employment, or on the ability to live independently, or on the quality of life of adults with DS. Readers are told that people with DS *can* achieve specific levels of

knowledge/social integration—e.g. "Individuals with DS can be employed competitively or in a workshop setting"—without any data on what proportion of this population *do* achieve these milestones; indeed, the lack of qualifiers could easily be interpreted as indicating that all individuals with DS achieve them.

Unfortunately, useful statistics on quality of life in DS are in short supply. As Robert Saul and Stephanie Meredith note, prospective parents want information about life outcomes—"what life is like for people living with specific genetic conditions in the real world" [30]. Qualitative studies that report the family and self-perceptions of individuals with DS, their siblings, and parents are helpful in this regard. The most frequently cited of these studies have been conducted in the U.S. by Brian Skotko and colleagues (for examples [31, 32]; see also [33-35]). However, as Skotko and other researchers explicitly recognize, the data from such studies, which typically recruit through DS advocacy organizations, is subject to non-response and selection bias. More generalizable population-based studies are rare and have focused much more on children and adolescents than adults; for exceptions, see [20, 36-38]. Thus, the authors of the NSGC practice guidelines acknowledge the "lack of published data regarding the long-term natural history for adults with Down syndrome" [28]. Clearly, much more research on quality of life, especially in adulthood, is needed for truly accurate and balanced counseling. Kolker and Burke also recommended that the decision to terminate a pregnancy be treated similarly, "with equally sympathetic women explaining how, upon learning they had fetuses with the same diagnosis, they reached opposite conclusions" [27]. Their proposals remain at least as germane now as they were then, acknowledging as they do that information in this domain is always from some perspective. Implementing them systematically would go a long way towards achieving genuine balance in prenatal counseling.

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**Author Contributions**

Diane Paul drafted the manuscript, which was edited by Ilana Löwy. The content of the manuscript is based on research conducted by both authors.

**Competing Interests**

The authors have declared that no competing interests exist.

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