

# Prenatal Screening for Fetal Aneuploidy: A Commentary by the Canadian Down Syndrome Society

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The Canadian Down Syndrome Society (CDSS) is Canada's national voice for people with Down syndrome. Our vision is of a proud Canada where all are welcome, where diversity is embraced, and where everyone's genes are valued equally. Our mission is to ensure equitable opportunities for Canadians with Down syndrome. This mission extends into the ethical implications of prenatal screening.

Two of our eleven-member board of directors have Down syndrome. We also benefit from the input of an advisory committee consisting of twelve adults with Down syndrome from across Canada. These self-advocates are university students, entrepreneurs, artists, employees, and spouses, who share our concern about the new recommendations from the Society of Obstetricians and Gynaecologists of Canada (SOGC)<sup>1</sup> and the implications of those recommendations for them and other people with disabilities.

The CDSS defines Down syndrome as a naturally occurring chromosomal arrangement that has always been a part of the human condition, seeing it as neither a disease nor a negative medical outcome of pregnancy. Down syndrome, in and of itself, does not require screening, treatment, or cure. The chromosomal arrangements that result in the Down syndrome phenotype are neither good nor bad per se and are given a value only through the social constructs, knowledge base, and belief systems that surround Down syndrome. Down syndrome is not a birth defect or an illness. A person with extra genetic material from the 21st chromosome may, however, be predisposed to certain illnesses and medical conditions that are also present in the general population. Extra genetic material from the 21st chromosome may also provide protection against other illnesses and medical conditions, such as solid tumours.<sup>2</sup>

Fair and equitable access to prenatal screening may provide Canadian women with opportunities to make the best decisions for themselves and their families in terms of their reproductive health; however, if the information presented is not value-neutral, non-directive, and non-prejudicial, the decision-making opportunity is flawed. To wit, the notion of “screening for genetic disorders” presupposes that Down syndrome is indeed a disorder, a presumption the CDSS vehemently contests.

The call to expand the scope of prenatal screening to include all pregnant women in Canada must be accompanied by a re-evaluation of the assumptions about and historical perceptions of citizens with Down syndrome. These assumptions and perceptions are perpetuated by language that characterizes Down syndrome as a negative medical outcome of pregnancy.<sup>3</sup>

To ensure the realization of the comprehensive program recommended, and not simply the technology of testing, it is important to separate cultural impressions of Down syndrome from the provision of fair and balanced information to Canadian women.<sup>4</sup> Discussion about the availability and implications of prenatal screening must be accompanied by fair, balanced, and value-neutral information about Down syndrome and the realities of life with a disability. To achieve this, health care providers need knowledge beyond that of screening modalities.

The CDSS applauds the SOGC for acknowledging the importance of and need for “informed consent” and “non-directive counselling” but offers a caution about the difficulty of achieving truly informed consent and of delivering non-directive information without providing a comprehensive education program in disability awareness. Medical professionals who discuss Down syndrome with prospective parents need to be well acquainted with the resources available to women and their families, including

information about genetic counselling services, parent support groups, early intervention and developmental clinics, and relevant specialists in their geographic area. If front line health care providers lack this information, telling a pregnant woman about a fetal Down syndrome karyotype is prejudicial and contrary to the SOGC's recommendation that "screening programs should show respect for the needs and quality of life of persons with disabilities." The CDSS advocates for a requirement that health care providers be educated about Down syndrome, a component missing from these guidelines that negates their good intentions. Further, to ensure an environment in which balanced and value-neutral decision making exists, CDSS believes increased funding for screening must be accompanied by appropriate funding to promote increased public awareness and to develop information materials that can be provided to pregnant women before they are given a *choice* to undergo testing or not.

Information about prenatal gender determination is generally readily available to prospective parents. However, in North America, it is generally not morally acceptable for them to seek this information for the purpose of gender selection. This practice with its potential ethical dilemma is intimately connected to the cultural and societal norms dictated by the ethnicity of any given community.<sup>5</sup> The CDSS cautions the medical community against distinguishing between the social pressures that may promote the desire for a child of a particular gender (and the actions associated with that desire) and the subtle but powerful messages that prospective parents receive about the potential quality of life of a child with Down syndrome.<sup>5</sup>

The College of Physicians and Surgeons of British Columbia supports the Commission on New Reproductive Technology, whose report states that the use of ultrasound for fetal gender determination for non-medical purposes prior to 20 weeks' gestation is unethical, even though in many other cultures this is normal and accepted practice.<sup>6</sup> We contend that neither the selective screening for gender nor the selective screening for Down syndrome is medically based, but rather that both are cultural in origin. Until Canadians, including health care professionals and prospective parents, are better informed about Down syndrome and the valuable contributions citizens with Down syndrome make to Canadian society, this culturally directed screening and subsequent terminations of fetuses with Down syndrome will continue.

Further, targeted testing and termination of pregnancies that will result in infants perceived to have a poor quality of life are generally accepted from a bio-ethical perspective. This perception is the result of social conditions.<sup>4</sup> The ethical implications of selective termination because of gender

and because of disability are similar. In Canadian society, women and men are understood to have an equitable quality of life, and, as a result, the screening and termination of pregnancies on the basis of gender preference are considered abhorrent. The consistent use of unbiased information and value-neutral language is important in influencing the prospective parents.

The power of words is profoundly evident in the language of pathology used in the medical context of describing Down syndrome. Even the premise of screening for Down syndrome itself, instead of the medical conditions typically associated with it, is a subtle but clear message about the lack of value placed on the lives of individuals with disabilities. This increases the difficulty of following the SOGC's recommendation no. 10, regarding showing respect for the needs and quality of life of persons with disabilities.<sup>1</sup>

"Discomfort with the use of stigmatizing labels was one of the primary reasons for SLANG (Sensitive Language and the New Genetics). We are not suggesting that it is possible to create a language that could be devoid of problems or misuse: *Rather we seek to raise the awareness of health professionals on the importance of language choice.*"<sup>3</sup> The practice of using value-neutral and person-first language, noting the chance or probability of having a baby with Down syndrome, rather than the risk of having a Down syndrome baby, in prenatal care and counselling is a critical part of preserving the right of women and families and citizens with disabilities to self-determination.<sup>3,4</sup>

Throughout the SOGC's Clinical Practice Guidelines and associated press releases there is abundant use of value-laden references to Down syndrome, including *risk*, *defect*, *disorder*, *abnormal*, and *anomaly*. Alternatives such as *chance* and *probability* in place of *risk* and *difference* in place of *abnormal* and *anomaly* are recommended in the literature.<sup>3,4</sup> Skotko states that "the word *risk*, by definition, predicts the likelihood of an undesired outcome. A non-directive statement would have used the word *chance*."<sup>4</sup> The CDSS promotes the use of the term *determination* rather than *diagnosis* regarding the presence of prenatal and postnatal indicators for Down syndrome as a value-neutral alternative.<sup>3,4</sup>

Although parents generally dream of having a perfect child, the reality of parenthood and the human condition guarantees the modification of that dream soon after birth. Resources, information, technology, and non-directive obstetrical care would be better directed to the natural and unavoidable risks of pregnancy, delivery, and early childhood development than to a targeted screening program if that program does not ensure balanced, unbiased, value-neutral information before options for testing are provided.

To dismiss the social construct surrounding targeted genetic screening for Down syndrome is, in essence, to dismiss the human condition. Information is never given or received in a vacuum.

We strongly recommend the inclusion of supplementary recommendations for patient and professional education. Physicians and other health care providers need to be much better informed about the reality of lives lived with Down syndrome.<sup>7,8</sup>

Recommendation no. 10 states that screening programs should show “respect for the needs and quality of life of persons with disabilities.” To facilitate that, there must also be a recommendation to provide current value-neutral information about Down syndrome. Also missing from the recommendations are (1) a provision for putting prospective parents in contact with support groups or others who are qualified to provide information about the reality of life with Down syndrome and (2) a commitment to ensuring that health care providers themselves are informed about living with Down syndrome.

We recognize that advances in medical technology have profound positive implications for prenatal care. The CDSS encourages ongoing and in-depth public discourse on the

ethical implications of genetic screening for Down syndrome and other disabilities.

*“Words are, of course, the most powerful drug used by mankind.”*<sup>9</sup>

## REFERENCES

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