

Canadian College of Medical Geneticists and Canadian Association of Genetic Counsellors commentary to proposed Ontario Bill 225 and Ontario Bill 304 (Harvey and Gurvir's Law 2020 and 2021, respectively)

The Canadian College of Medical Geneticists (CCMG) and the Canadian Association of Genetic Counsellors (CAGC) wish to comment in response to proposed Bill 225: An Act to amend the *Regulated Health Professions Act, 1991* respecting the provision of information respecting Down syndrome by regulated health professionals and to proposed Bill 304: An Act to amend the *Regulated Health Professions Act, 1991* respecting the provision of information about Down syndrome to expectant parents, regulated health professionals and the public (Harvey and Gurvir's Law 2020 and 2021, respectively). Bill 225 and Bill 304 were introduced in the Legislative Assembly of Ontario (OLA) on November 2, 2020 and June 2, 2021, respectively (first readings carried). While we understand that this specific proposed legislation is not under active consideration by the OLA, its introduction has prompted our organizations to provide our perspective on this and all substantively similar legislation.

We would like to start by recognizing that this proposed legislation and the experiences of those who brought it forward identify important concerns about the current availability and provision of accurate, balanced information about the range of intellectual, developmental, medical, and psychosocial outcomes for individuals with Down syndrome. We wish to acknowledge these experiences. We support that all health care providers, regardless of specialty or practice setting, who communicate a likely or confirmed diagnosis of Down syndrome to parents or expectant parents have a responsibility to deliver accurate information and to engage in continuous education. We express hope that other individuals and families in Ontario and across Canada do not have negative experiences over the course of their journey through the healthcare system.

Before outlining our comments, please allow us to introduce ourselves. Medical geneticists are medical doctors and laboratory scientists who are specialists in medical or laboratory genetics, respectively, with expertise in the diagnosis and treatment of genetic conditions. Genetic counsellors are health care providers with specialized training in both medical genetics and counselling who typically hold a Master's degree in genetic counselling and Board certification. We often work together in medical genetics clinics across Ontario and Canada, or with other care providers such as obstetricians, to provide patient-centred, evidence-based care to patients and families about genetic conditions. The CCMG represents almost 300 medical geneticists. The CAGC represents over 350 genetic counsellors.

Up-to-date, evidence-based information about Down syndrome

Bill 225 and Bill 304 propose that Ministry-approved up-to-date, evidence-based written information relating to Down syndrome be made available to expectant parents and to the public.

The CCMG and CAGC strongly support the provision of up-to-date, evidence-based written and verbal information to the public and to expectant or current parents. A central tenet of our roles as Genetics professionals is to communicate current and reliable information about genetic conditions and testing options in a way that is meaningful to individuals or families navigating complex medical information and testing, with the appropriate use of medical interpreters, counselling aids, and written materials, as needed¹⁻⁶. The aim is to facilitate informed decision-making in a supportive, non-directive setting that is respectful of the ethnocultural and social values unique to each person or family¹⁻⁶. We strongly believe that individuals or families can make autonomous decisions based on accurate and complete information¹⁻⁶, which includes understanding the full spectrum of a diagnosis.

We do, however, believe that the provision of up-to-date information can be achieved without being a legislated requirement. Such legislation would represent a deviation from the Canadian health care model, where recommendations and standards are set out by our professional bodies¹⁻¹¹. It would demonstrate

exceptionalism towards Down syndrome, and would neglect to address the thousands of other genetic conditions diagnosed prenatally where we would like to provide the same compassionate, patient-centred, and up-to-date expert opinion to individuals and families. Effectively conveying a diagnosis of Down syndrome involves more than reviewing a checklist or a fact sheet of Ministry-approved information; it involves a nuanced and thorough discussion - sometimes multiple discussions at follow-up visits as needed - founded in an assessment of the unique informational and emotional needs of expectant or current parents and on a non-directive approach to counselling. We worry that written material alone may not be inclusive of all cultures, languages, and literacy levels, but appreciate the merit of written materials in many contexts. We will continue to communicate reliable, supportive, and current information as a central component of patient-centred care and our own codes of ethics. We are available as a resource to patients, to health care providers, and to the general public with the aim to facilitate the accurate dissemination of information about Down syndrome and other genetic conditions.

Mandatory 48-hour waiting period after diagnosis

Bill 225 additionally proposes a mandatory waiting period of 48 hours after a prenatal diagnosis of Down syndrome before health care professionals can recommend further tests or treatment relating to this diagnosis, unless explicitly requested by the patient or unless the health care provider deems the testing or treatment necessary during the 48 hour period. We note that the more recent iteration of Harvey and Gurvir's Law (2021, Bill 304) does not include this requirement.

The CCMG and CAGC do not think a mandated 48-hour waiting period following the disclosure of a prenatal diagnosis of Down syndrome - or similarly of a high-risk non-invasive prenatal test (NIPT) result – to recommend further tests or treatments is in the best interests of all patients/families. A mandated waiting period would undermine a health care provider's ability to have transparent and truthful discussions with their patients. It may jeopardize the trust and rapport that are central to the health care provider-patient relationship. We are concerned that 48 hours during which options for patients are not provided may jeopardize the provision of truly informed consent, may delay time-sensitive care, and may cause additional anxiety, as a mandated waiting period will add to the already inherent time lag between when patients receive a positive prenatal screen result and when they receive a definitive diagnosis. Ideally, discussions surrounding the risks, benefits, and outcomes of a test – including a discussion of all possible follow-up tests or treatment - should occur *before* an individual undertakes testing as part of the informed consent process. For those who may have already made a decision following pre-test counselling, an extra delay may be particularly harmful. Finally, further tests or treatments should be presented as *options* rather than *recommendations* in a non-directive, shared decision-making model that considers unique patient preferences, beliefs, and life circumstances, based on the provision of complete and timely information. The American College of Medical Genetics and Genomics (ACMG) has stated a similar position on legislation in the United States¹².

Commitment to partner in promoting excellent care

Ensuring positive and supportive experiences for individuals or families facing decisions about Down syndrome screening and testing is a goal that we share with those who have brought these Bills forward. Our organizations would welcome the opportunity to contribute to the development and dissemination of up-to-date, accurate educational resources with relevant stakeholders including parents, advocacy groups, and other medical professional colleagues and associations¹³⁻¹⁴. We also commit to supporting initiatives that promote increased education to health care providers about prenatal screening tests, prenatal diagnosis, Down syndrome and other genetic conditions. Finally, the CCMG and CAGC would welcome the opportunity to provide further expert opinion and guidance prior to the consideration of the same or similar legislation by the Legislative Assembly of Ontario, or by any other Canadian legislative body.

This Commentary is respectfully submitted by the Canadian College of Medical Geneticists and the Canadian Association of Genetic Counsellors.

SUMMARY OF KEY POINTS

- The CCMG and CAGC strongly support the provision of up-to-date, evidence-based written and verbal information to the public and to expectant or current parents
- However, we believe that the provision of up-to-date information can be achieved without being a legislated requirement
- We do not think a mandated 48-hour waiting period following the disclosure of a prenatal diagnosis of Down syndrome - or similarly of a high-risk non-invasive prenatal test (NIPT) result – to recommend further tests or treatments is in the best interests of all patients/families
- We commit to supporting initiatives that promote increased education to health care providers about prenatal screening tests, prenatal diagnosis, Down syndrome and other genetic conditions
- We will continue to communicate reliable, supportive, and current information as a central component of patient-centred care
- We would welcome the opportunity to provide further expert opinion and guidance prior to the consideration of the same or similar legislation by the Legislative Assembly of Ontario, or by any other Canadian legislative body

BACKGROUND INFORMATION ABOUT PRENATAL SCREENING AND PRENATAL DIAGNOSIS IN ONTARIO

According to professional clinical practice guidelines, pregnant individuals in Canada should be offered the option of prenatal screening for common chromosome conditions, including Down syndrome, through an informed counselling process⁹. **Prenatal screening aims to identify whether there is a higher chance in a pregnancy of having a baby with Down syndrome compared to a pregnant individual's baseline age-related chance.** There are several prenatal screening modalities available. Options include multiple marker screening (MMS) - most commonly first trimester screening (FTS) - to analyze ultrasound and/or biochemical (fetal/placental) markers, as well as noninvasive prenatal testing (NIPT) that analyzes circulating fragments of placental cell-free DNA (cfDNA) in a pregnant individual's blood. Prenatal screening methods vary significantly in their ability to detect Down syndrome, and in the probability that a positive screen result represents a true diagnosis. FTS results are most commonly available at the end of the first trimester of pregnancy (approximately 12-14 weeks gestation). NIPT can be arranged after 9 or 10 weeks gestation, and results typically take 1-2 weeks¹⁵. Prenatal screening services in Ontario are coordinated by Prenatal Screening Ontario (PSO) (see www.prenatalscreeningontario.ca).

Importantly, neither MMS nor NIPT is a diagnostic test: While NIPT identifies >99% of pregnancies with Down syndrome, its positive predictive value (PPV) - meaning the likelihood that a high risk NIPT result truly represents a diagnosis of Down syndrome – depends on several factors but is approximately 90% on average. In other words, **up to 1 in 10 high risk NIPT results for Down syndrome do not represent a true diagnosis.** Therefore, professional associations including the Society of Obstetricians and Gynaecologists of Canada (SOGC) and CCMG emphasize that **irrevocable pregnancy decisions should not be made on the basis of a screening result alone**⁹.

Two invasive procedures called chorionic villus sampling (CVS) and amniocentesis can confirm a diagnosis of Down syndrome prenatally. These procedures are typically only arranged following a detailed consultation in a medical genetics clinic, maternal fetal medicine (MFM), or perinatology/high risk obstetrical service. **Offer of a referral to one of these specialist services for a discussion of all options - including the option of no further testing – is appropriate for pregnant individuals who have received a positive prenatal screen result.** Prenatal diagnosis is offered only after a detailed discussion of all options, including risks, possible outcomes, and alternatives to testing.

Authorship:

This statement was produced by a joint working group and has been endorsed by the CCMG and CAGC Boards of Directors

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