

Genetics and Paediatric Health: Section 6

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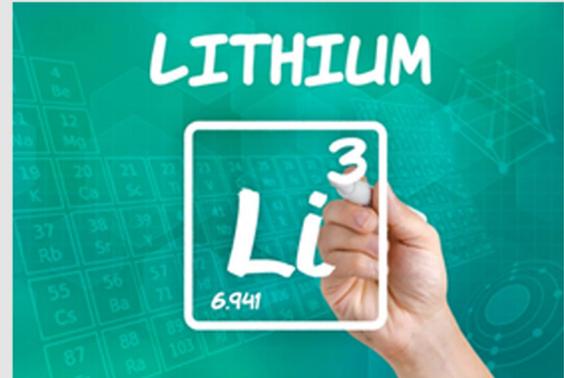
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6.1.1 Introduction to Teratogens



What do recreational drugs, lithium, phenylalanine, and chicken pox have in common?



They are all examples of possible teratogens.



Image credit: Shutterstock

The human epigenome responds to the environment in a very dynamic fashion. For example, toxic exposure, stress, diet, and other factors influence chemical “switches” that control gene expression. Teratogens are one of these factors.

A teratogen is a substance known to cause birth defects following exposure during pregnancy. Some teratogens can be drugs (e.g., prescription drugs such as lithium or epilepsy medication or recreational drugs). Certain infections, such as rubella (German Measles) or chicken pox can also be teratogens. The mother can also unknowingly introduce the fetus to teratogens in the womb. For example, in the case of PKU, a mother with PKU herself, who does not follow the prescribed diet, has very high levels of phenylalanine that passes to her fetus through placental circulation and leads to congenital anomalies in the fetus, such as congenital heart disease.

It is often geneticists and genetic counsellors who provide counselling for mothers and infants exposed to teratogens.





6.1.2 Alcohol



Image credit: Best Start: http://www.beststart.org/resources/alc_reduction/pdf/static_clings_bi_lr.pdf

Alcohol is a common teratogen. Alcohol use during pregnancy can adversely affect the unborn baby. There are many factors that influence this effect, including the amount of alcohol ingested over time and differences in the way the mother metabolizes alcohol. There is also evidence that variations in a person's genetic makeup can affect the baby's susceptibility to alcohol while in utero.¹

The effects of alcohol exposure during pregnancy are considered on a spectrum, and thus are called fetal alcohol spectrum disorder (FASD). These effects can include physical, mental, behavioural, and learning disabilities, as well as cognitive, emotional, and behavioural issues.²

The medical diagnoses of FASD include Fetal Alcohol Syndrome (FAS), Partial FASD (pFAS), and Alcohol-Related Neurodevelopmental Disorder (ARND).³ The diagnosis is always related to restriction in growth, changes in facial features, problems with the central nervous system, brain damage, and prenatal exposure to alcohol.⁴

¹ Reynolds JN, Weingbert J, Clarren S, Beaulieu C, Rasmussen C, Kobor M, et al. Fetal alcohol spectrum disorders: gene-environment interactions, predictive biomarkers, and the relationship between structural alterations in the brain and functional outcomes. *Semin Pediatr Neurol*. 2011;18(10):49–55

² Chudley AE, Conry J, Cook JL, Loock C, Rosales T, LeBlanc N. Fetal alcohol spectrum disorder: Canadian guidelines for diagnosis. *CMAJ*. 2005;172(5 suppl)

³ Public Health Agency of Canada, Fetal Alcohol Spectrum Disorder (FASD). <http://www.phac-aspc.gc.ca/hp-ps/dca-dea/prog-ini/fasd-etcaf/index-eng.php>

⁴ Carson G, Cox LV, Crane J, Croeau P, Graves L, Kluka S, et al. Alcohol Use and Pregnancy Consensus Clinical Guidelines. SOGC Clinical Practice Guideline. *JOGC*. 2010; 32(8). <http://sogc.org/wp-content/uploads/2013/01/qui245CPG1008E.pdf>

MOTHERISK, at the Hospital for Sick Children, is an excellent resource for more information on alcohol and pregnancy, and the effects of other drugs and substances during pregnancy. MOTHERISK offers The Alcohol and Substance Use Helpline (1-877-327-4636), which provides information and counselling to pregnant and breastfeeding women, their families, and healthcare providers.





6.1.3 Fetal Alcohol Spectrum Disorder – The Facts

FETAL ALCOHOL SYNDROME

Affects
1 to 3 / 1,000
Babies born in Canada.

FETAL ALCOHOL SPECTRUM DISORDER

Affects **10/ 1,000**
babies born in Canada.
This can be as high as **190/1,000 live births** in
some Canadian communities.

Graphic created by CICH using data from Robinson GC, Conry JL, Conry RF. Clinical profile and prevalence of fetal alcohol syndrome in an isolated community in British Columbia. CMAJ 1987;137:203–7.

It is estimated that 1 to 3 per 1,000 babies born in Canada have fetal alcohol syndrome (FAS). Fetal alcohol spectrum disorder (FASD) is estimated to affect approximately 1% of the population. Some communities in Canada report prevalence rates as high as 190 per 1,000 live births.¹

¹ Robinson GC, Conry JL, Conry RF. Clinical profile and prevalence of fetal alcohol syndrome in an isolated community in British Columbia. CMAJ. 1987;137:203–7

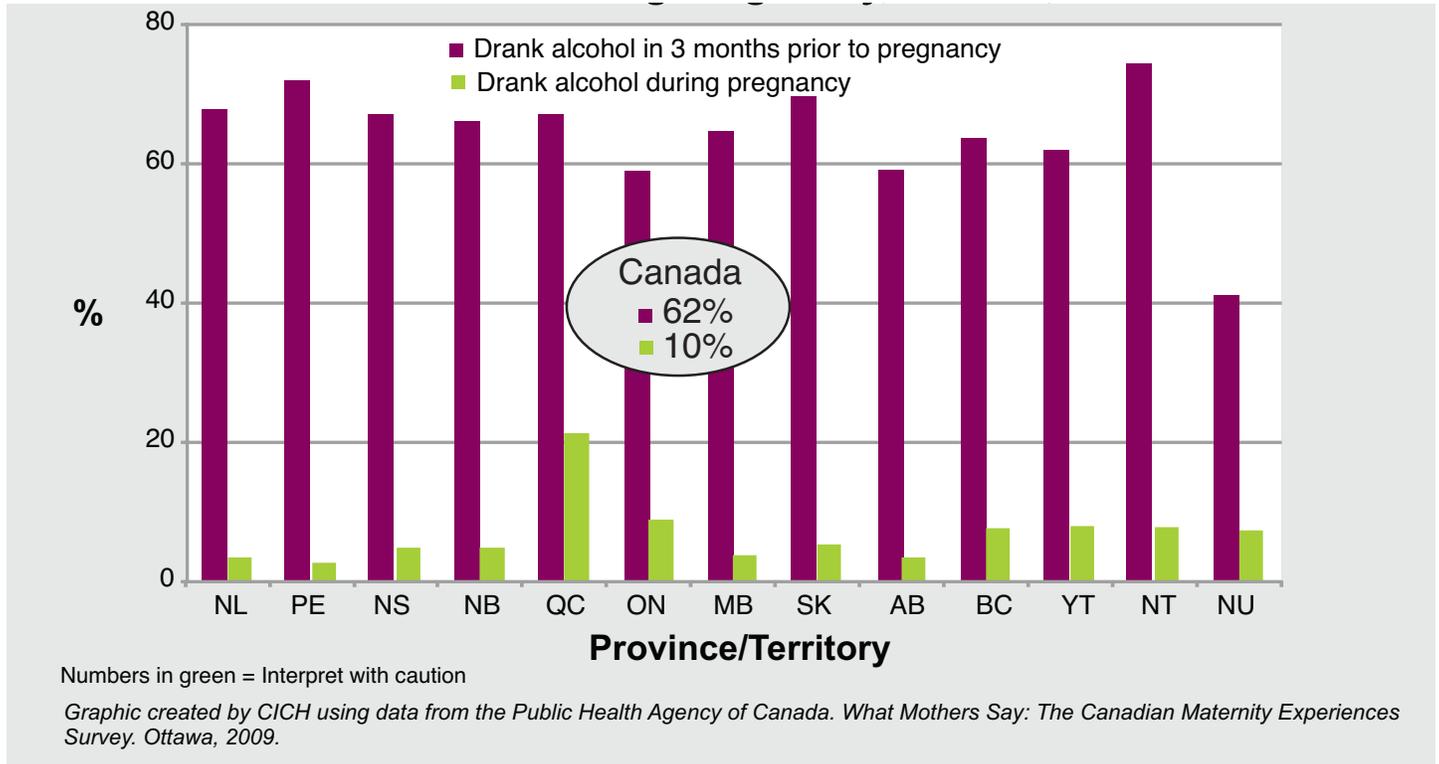
Implications

FASD cannot be cured. It can have lifelong effects on individuals, their families, and their communities. There is a need to develop a better understanding of risk factors associated with FASD.





6.1.4 Fetal Alcohol Spectrum Disorder – Alcohol Use During Pregnancy, Canada, 2009



According to a national survey of new mothers, 62.4% of women reported drinking alcohol during the three months before their pregnancy. However, only 10.5% of women reported consuming alcohol during pregnancy, and that number varied by province and territory. Less than one percent (0.7%) of mothers reported drinking frequently. Furthermore, 11% indicated they had engaged in binge drinking before realizing they were pregnant.¹

¹ Public Health Agency of Canada. What Mothers Say: The Canadian Maternity Experiences Survey. Ottawa: Public Health Agency of Canada; 2009





6.1.5 Fetal Alcohol Spectrum Disorder – Primary Prevention



FASD is the leading cause of preventable developmental disability among Canadians. Because health behaviours must be considered within the greater context of the lives of women and their families, women require a variety of support, education, and policy approaches to enable them to maintain their health. A number of national groups in Canada have endorsed evidence-based practice guidelines – Alcohol Use and Pregnancy Consensus Clinical Guidelines – to help practitioners support women around alcohol use.¹

¹ Endorsed by: The Society of Obstetricians and Gynaecologists of Canada; Motherisk; The College of Family Physicians of Canada; Canadian Association of Midwives; Association of Obstetricians and Gynecologists of Quebec; Federation of Medical Women of Canada; Society of Rural Physicians of Canada; Canadian Association of Perinatal and Women's Health Nurses.

Alcohol Use and Pregnancy: Consensus Clinical Guidelines

- Universal screening for alcohol consumption should be done periodically for all pregnant women and women of child-bearing age.
- Healthcare providers should create a safe environment for women to report alcohol consumption.
- The public should be informed that alcohol screening and support for women at risk is part of routine women's healthcare.
- Healthcare providers should be aware of the risk factors associated with alcohol use in women of reproductive age.
- Brief interventions are effective and should be provided by healthcare providers for women with at-risk drinking.
- If a woman continues to use alcohol during pregnancy, harm reduction/treatment strategies should be encouraged.
- Pregnant women should be given priority access to withdrawal management and treatment.
- Healthcare providers should advise women that low-level consumption of alcohol in early pregnancy is not an indication for termination of pregnancy.

Source: Carson G, Cox LV, Crane J, Croeau P, Graves L, Kluka S, et al. Alcohol Use and Pregnancy Consensus Clinical Guidelines. SOGC Clinical Practice Guideline. JOGC. 2010;32(8). <http://sogc.org/wp-content/uploads/2013/01/gui245CPG1008E.pdf>





6.1.6 Fetal Alcohol Spectrum Disorder – Primary Prevention



Graphic created by CICH using a graphic from Best Start, http://www.beststart.org/resources/alc_reduction/index.html

It is essential that women and families have accurate information about drinking alcohol during pregnancy.

Some Myths and Facts about Alcohol and Substance Use

MYTH: Alcohol or drugs taken after the first trimester will not affect the unborn baby.

FACT: Most organ development is completed a few weeks after the first trimester. Brain development continues throughout pregnancy and after birth. Exposure to substances any time in the pregnancy can affect the baby's brain.

MYTH: One drink in pregnancy is enough to harm the unborn baby.

FACT: A safe amount of alcohol in pregnancy is not known. It is unlikely, though, that a single drink before a mother knew she was pregnant could damage her unborn baby.

MYTH: There is no hope for a baby exposed to heavy drug and alcohol use.

FACT: There is always hope. Drug and alcohol use in pregnancy affects each baby differently.

Source: MOTHERISK, <http://www.motherisk.org/women/alcohol.jsp#two>

