Genetic counsellors often work as part of a multidisciplinary team that may include geneticists, paediatricians, nurses, or other specialists such as OB-GYNs. The family below was seen in such a clinic. Often families who are seen in a Genetics clinic have seen a multitude of specialists and may go undiagnosed for many years; this is often referred to as the “diagnostic odyssey”. The story below illustrates one family’s experience. Genetic counsellors help families like this understand and adapt to the medical, psychological, and familial implications of genetic conditions throughout the diagnostic odyssey.

For the longest time, our son Liam was undiagnosed. The medical community (dozens of doctors and specialists) could only shake their heads, and gave him the diagnosis of “globally delayed of undiagnosed etiology with autistic tendencies and a seizure disorder”. No one had any answers or could give us any hope of a diagnosis. No one, that is, until we met a geneticist—Dr. Walia. Dr. Walia believed that our son had a genetic disorder.

Every year he would send Liam’s samples for different genetic testing. The year Liam turned 17, the government opened up a pocket of funding to do some more genetic testing on a few individuals. Dr. Walia selected Liam to be part of this project. This is the year Liam was finally diagnosed with KIAA2022.

I used to tell people that Liam was “one of a kind”, and I now know that to be true. He currently is the only person in Canada with this diagnosis. Up until the time of diagnosis, my husband was riddled with fear and guilt.
Did something happen to Liam when he was in the care of his biological mother? (I am his adoptive mother.) Could he have somehow prevented what was wrong with Liam? Would this disability show itself in our future children together? (Liam now has a younger brother and sister, and both are healthy.) When we finally got the diagnosis, all the fears and feelings of guilt he had, were put to rest. With that relief also came the realization that we had spent over a dozen years fighting and advocating for someone to give us an answer! Since the diagnosis, I have been able to connect with 3 other families around the world whose sons are also affected by KIAA2022. We have been able to share stories, suggestions, videos and pictures. We “get” what each other is going through. The similarities in our sons is amazing, to the point that Liam looks and behaves just like one of the other boys. But how many other undiagnosed children (and parents) are out there, all alone, and wondering? Words cannot express how great it is to no longer feel like we are alone with a child such as ours. We now have some support, even though it is only from a few other diagnosed families, who live halfway around the world.

Kirsti Bakker
Liam’s Adoptive Mom

In 2014, the CAGC proudly supported the Federal Court challenge by the Children’s Hospital of Eastern Ontario (CHEO) launched against Transgenomics, Inc., the owner of five gene patents related to the potentially deadly Long QT syndrome.

On March 9, 2016 CHEO announced a deal that ensures Canadian public sector hospitals and laboratories the right to test for Long QT syndrome for Canadian patients.

What’s more, it sets a precedent that will help address the issue of gene patents more broadly in Canadian health care.

In the settlement, the patent holder Transgenomic has agreed to provide CHEO and all other Canadian public sector hospitals and laboratories the right to test Canadians for Long QT syndrome on a not-for-profit basis. The deal defines a pathway for all public Canadian hospitals and labs to conduct genetic testing without legal roadblocks from gene patents.

Behind the scenes, the CHEO team included Geneticists, Laboratory Scientists, pro-bono lawyers, Hospital Executives and a Genetic Counsellor who worked collaboratively over several years to make March 9th a great day for Canadian patients and their families.

To learn more about this achievement, please visit: www.cheo.on.ca/en/gene-patents

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CHECK OUT

LEXIGENE®

a bilingual tool that allows an individual to search for genetics-related terms in either French or English and find the equivalent term in the other language. This online tool boasts over 3500 translated terms and is intended for use by genetic counsellors, geneticists, and others who work in the field of genetics in both English and French.
Although much progress has been made, Canada remains the only G8 country that does not have laws to protect its citizens against genetic discrimination based on personal genetic testing results or family history of a genetic condition. In 2012, the CAGC released a Position Statement on Genetic Discrimination to advocate for the protection of Canadians against genetic discrimination (full statement).

On April 14th, 2016, the Senate passed The Genetic Non-Discrimination Act, also known as Bill S-201, “An Act to prohibit and prevent genetic discrimination”. The bill proposes protection for Canadian citizens against genetic discrimination by prohibiting any person from requiring an individual to undergo a genetic test or disclose the results of a genetic test, and by amending the Canadian Human Rights Act to prohibit discrimination on the ground of genetic testing results or family history of a condition.

Once a bill is passed by the Senate, in order for it to be enacted into law, the bill must go through three readings in the House of Commons. The first reading in the House of Commons took place in May 2016 and the second reading occurred in September. The bill is currently being considered in the House of Commons. Debate on the second reading of the bill is scheduled to continue on October 25. If a majority vote in support of the bill after the debate, it will proceed to committee stage for further review and testimony, before passage to a final reading and vote.

For more information about Bill S-201 please see the full text of the bill. Individuals who wish to provide support for Bill S-201 can do so by contacting their local MP.

**WHAT IS A GENETIC COUNSELLOR?**

Health professional with specialized training and experience in the areas of medical genetics and counselling who:

- Provides individuals and families with information on the nature, inheritance, and implications of genetic conditions to help them make informed medical and personal decisions
- Helps people understand and adapt to genetic conditions including seeking appropriate medical management and surveillance
- Provides supportive counselling and connects people to resources

**FIND A GENETICS CLINIC NEAR YOU!**

Click here for more information.
My name is Heather. I am a wife, mom of two cute kids and I work full time at a national charity, sit on school parent council, and am a Girl Guide leader. I have been fairly healthy all my life. My perspective on my health changed when I was 29. In my mid-twenties, I had a colonoscopy that revealed I have polyps but there was not much concern. Years later, when I met my new nurse practitioner, she suggested that I have a follow up colonoscopy. The colonoscopy revealed that there were more unusual polyps and a sample was to be sent to the pathologist. After a couple of months, I heard back that I was being referred to a Genetic Counsellor to follow up.

I waited for my appointment with the Genetic department, trying to be patient. Finally, the day came. I met my Genetics Doctor and Genetic Counsellor. They explained why I was there, how genetic testing may impact my life, and explained genetics in a way that I could somewhat understand. It was overwhelming to be told that I may have a genetic mutation, but it was comforting to have these two professionals asking how I am feeling along the way. My genetic testing revealed that I have JPS, aka Juvenile Polyposis Syndrome. Neither myself or my family doctor had heard of this rare condition. I also learned that with my SMAD4 mutation, I may have another rare genetic condition called HHT, aka Hereditary Hemorrhagic Telangiectasia. Follow up tests were encouraged.

What did this mean? Learning that I have increased risks for certain types of cancers left me initially shocked, upset, and scared. Not only did this affect me, but it affects my husband, two young kids, parents, siblings, and other family members. When I think about it, genetic testing is difficult because there are so many factors – how will you feel about yourself, how will others feel when they know this about you, how it affects your life (work, insurance, etc.), what tests and screenings you need to have done, which doctor to see, how it impacts some of your future decisions...

I became focused on learning about JPS and HHT. The more I understood about these two conditions, the better I was able to work through these emotions. I choose to see the positives in this diagnosis. My husband, in-laws, parents, and friends support me when I feel overwhelmed. I choose to appreciate how my young daughter and mom and possibly other family members can be screened too. Luckily, I have a great employer who is supportive and understanding of medical appointments. Also, thank goodness I have life insurance in place (thank you mom and dad!), as these things were now more complicated. Through Mount Sinai’s genetic registry, I was connected to someone else who has a similar diagnosis. This has been a rewarding experience for me to work through this diagnosis. However, the most comforting of all is that I am receiving consistent and wonderful health care from an expanded team of doctors, specialists, and the Genetics team. These professionals are looking out for my wellbeing today and in the future.

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**Education**

**How to Become a GC**

Master of Science degree in Genetic Counselling obtained from a certified training program (Canada offers 5 programs, there are also programs in the United States and abroad). Programs are 2 years in length and include a capstone/thesis project, coursework in genetics and counselling theory, and clinical rotations.

Pre-requisites* include:

- Undergraduate degree in related field + related coursework in genetics, biology, psychology, etc.
- Volunteer experience in counselling eg. distress or crisis line, planned parenthood, rape crisis centre
- Exposure to genetics clinic eg. volunteer or work experience

* For an up-to-date list of available programs and pre-requisites, please refer to the [CAGC website](https://cagc.ca) or the [ABGC website](http://abgc.ca).
The Canadian Association of Genetic Counsellors (CAGC) Certification Board is committed to ensuring that members of the Canadian public are receiving genetic counselling services from health care professionals with a standard level of knowledge, skills, attitudes, and judgement. Genetic counsellors granted the credential CCGC (Canadian Certified Genetic Counsellor) and/or its French equivalent CGAC (Conseiller(ère) en génétique agréé(e) du Canada), have demonstrated these standard knowledge and practice competencies.

Look for the CCGC – Canadian Certified Genetic Counsellor – designation for quality and competence in genetic counselling.

- Although my life as a busy 31-year-old mom of two, working full time and volunteering when I can, now includes colonoscopies, CT scans, bubble echo tests, MRI, ultrasounds, video capsule endoscopy, etc. I am glad that in Canada, we have genetic doctors and counsellors here to help. I am thankful for our healthcare system that has made this testing and all my doctor support possible.

  Heather Brown

**CAGC ANNUAL EDUCATION CONFERENCE 2016**

This year’s Annual Education Conference (AEC) will be held November 3rd to 5th, 2016, in Montreal, Quebec. The conference will include a short course featuring well-known experts on ethics and policy in the genomic era, plenary sessions with updates on adult neurogenetics, CRISPR technology and treatment of genetic conditions, among many others! Anyone interested is welcome to attend! Please visit the conference website (www.cagcconference.ca/2016) for program and registration information. You can follow the conference on Twitter at #CAGC2016.

**JOIN US FOR GENETIC COUNSELLING AWARENESS WEEK 2016!**

What is genetic counselling? Since many people remain unaware of genetic counselling and the role that genetic counsellors have in helping with patient care, the GCAW is a movement by the CAGC to promote awareness of the profession! This year the 7th annual GCAW will be held from November 20th to 26th, 2016. For more information check out the CAGC’s Facebook page, Twitter handle and website for more information.