HOUSE OF COMMONS
CHAMBRE DES COMMUNES
CANADA

Standing Committee on Health

HESA • NUMBER 086 • 1st SESSION • 41st PARLIAMENT

EVIDENCE

Tuesday, May 7, 2013

Chair

Mrs. Joy Smith

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● (1530)

[*English*]

The Chair (Mrs. Joy Smith (Kildonan—St. Paul, CPC)): Good afternoon.

Welcome to the health committee and to the technological innovation study.

We're fortunate today to have as one of our witnesses, Ms. Jacquie Micallef, manager, member relations, Policy and Partnerships for Neurological Health Charities Canada.

Welcome.

We have via teleconference, Dr. Allan Micheil Innes, national coordinator for Orphanet Canada.

We're very happy that you're both here.

The procedure is that there will be two 10-minute presentations, followed by our Qs and As.

Dr. Innes, would you be so kind as to give us your 10-minute presentation.

It's hard with a video conference, but to keep you within the 10 minutes, I'll try to send you a signal when the end of the 10 minutes is coming up.

Dr. Allan Micheil Innes (National Coordinator, Orphanet Canada): Thank you, Madam Chair.

My name is Micheil Innes and it is an honour to address the committee today.

I'm a medical geneticist practising at the Alberta Children's Hospital in Calgary and at the University of Calgary. I'm also the national coordinator for Orphanet Canada, which I will discuss.

Today, I would like to emphasize the importance of securing an accurate diagnosis for all patients and families with rare diseases, with the goal of improving their health and wellness, as well as the current barriers that exist to achieving this goal.

As you have likely already heard, a rare disease is defined as one that affects one person out of 2,000. There are over 7,000 known rare diseases, which affect an estimated one in 12 Canadians. A disproportionate number of the nearly three million Canadians affected with rare diseases are children. As such, rare diseases affect a significant proportion of Canadian families. Most, but not all, rare diseases are genetic. Individuals with rare diseases suffer increased morbidity and mortality. As many as 30% of infants with a genetic disorder die in their first year of life. Children with rare diseases have more hospital admissions, with longer and more costly stays. This trend continues into adulthood, as older patients with rare diseases also have a disproportionate number of hospital admissions and costs. Hospital costs are only one factor to consider, as both outpatient medical and non-medical costs are also significant for the patient, their families, and the extended community. Patients with a rare disease face significant challenges, ranging from receiving a correct diagnosis to the availability of treatment and care.

The Chair: Excuse me, Dr. Innes, but our translators can't keep up with you.

Ms. Davies said she can't keep up with you in English. Our translators are having some difficulty translating your wonderful presentation.

Could you slow down a bit so the translators, and Ms. Davies, can pick it up?

Dr. Allan Micheil Innes: Absolutely. My apologies.

There are many critical reasons to secure a correct underlying diagnosis for patients and their families. First and foremost—

● (1535)

The Chair: Could you slow down a little bit because we want to hear everything you say. I'll give you an extra minute. You're still racing, so we can't keep up to you, doctor. You're way ahead of us.

Can you do that?

Dr. Allan Micheil Innes: Yes, I'm sorry.

I'm obviously finding it a little bit difficult from a distance here.

The Chair: I know.

Your material is exquisite. You're just brilliant.

Go ahead so we can hear you.

Dr. Allan Micheil Innes: Okay, thank you.

Patients or their parents deserve an answer as to their underlying condition. One can only imagine the significant guilt carried by parents of a child with an undiagnosed condition.

A diagnosis is the first step to understanding the natural history and prognosis of the disorder. Receiving an appropriate diagnosis can secure access to services in the school or community, appropriate medical surveillance, and on some occasions an underlying treatment. A diagnosis allows patients the opportunity to reach out to similarly affected individuals locally, nationally, or internationally through the form of patient support organizations. Finally, for genetic conditions, a correct underlying diagnosis can help clarify genetic counselling for the patient and other family members.

Orphanet is the world's online reference portal for information on rare disease. The portal is accessed internationally more than 20,000 times a day. Particularly for individuals diagnosed with a rare disease, it represents a comprehensive database of information, which can contribute to improvements in the diagnosis, care, and treatment of patients with a rare disease.

With support from the Canadian Institutes of Health Research, CIHR, Orphanet Canada with Canadian-specific content is now available. It provides Canadians with access to services in French and English related to rare disease, including an inventory of orphan drugs, a directory of services, clinics, laboratories, research projects, registries, etc. Canada was the first country outside Europe to formally join Orphanet, although in the past year Orphanet has become increasingly global with participation from Japan and Australia among others.

Since our formal launch, announced by the minister in October 2012, we have contacted over 350 stakeholders, and currently have validated information on over 70 such groups on our site. It is our desire that Orphanet increasingly be recognized in Canada by patients and physicians alike as a valuable early and ongoing resource for those with a rare disease.

However, even this basic information is not available to those individuals with a rare disease that has not been diagnosed. It's long been recognized that even in the best equipped medical genetics clinics in Canada or worldwide, more than 50% of patients do not receive a correct underlying diagnosis. There are many reasons for this, including the fact that the condition may be so rare that it has not even been discovered yet, or that the patient's presentation is atypical in some way, so that the diagnosis is not obvious.

Our current approach to genetic tests relies heavily on arriving at a best guess clinical diagnosis and then arranging specific testing for one of the over 20,000 genes in the human genome, with each test currently costing $1,000 to $3,000.

Internationally, there are currently about 2,500 genes for which testing is available, and of these, about 150 tests are available in Canada. As a result, provinces spend a significant amount of money, $18 million in Ontario alone in 2011, for genetic testing done internationally. Each province has developed its own approach to the issue, and access to such testing is quite asymmetric nationally. T

This “diagnostic odyssey”, which refers to the process, often takes years, and the cost for many patients is in excess of $10,000 to arrive at a diagnosis.

Within the last few years a new technology, next generation sequencing, has made it possible to sequence the entire genetic code of an individual in a more timely and cost-effective manner. It can be viewed as 22,000 genetic tests at once.

This technology has been leveraged very successfully by a recent Canadian consortium funded by Genome Canada and the CIHR, entitled FORGE Canada. Through remarkable collaboration of all genetic centres in Canada, this group was able to identify over 180 rare pediatric genetic disorders affecting hundreds of children in Canada and internationally. We were able to identify the genetic basis for 77 of these, and in the process to discover 45 new genes. This shows the value of this testing and collaboration in reaching a diagnosis for families.

These tests are available on a research basis at a cost of $2,000, and now commercially in the U.S. at a cost of approximately $10,000, but these costs should drop in the future. This testing should benefit thousands of Canadians moving forward. However, implementation of these tests clinically will not be trivial. It will require investing in infrastructure, developing approaches to data management and sharing, dealing with unexpected or incidental findings while remembering that we're sequencing a patient's entire genetic code to arrive at a specific diagnosis, and educating countless Canadian physicians and health care providers about these new technologies and their implications.

Of course arriving at a specific diagnosis is also critical in the selection of therapy. Of the approximately 3,500 known genetic rare diseases, only about 200 therapies are currently available. Health Canada's modern framework for the development of orphan drugs in Canada is certainly welcome and essential.

We should not forget that discoveries of drugs with major importance for the general population have often been made by first studying patients with a rare disease.

● (1540)

One such example is the development of the now widely prescribed statin drugs for high cholesterol, which were first studied and developed for patients with rare genetic forms of high cholesterol.

We often hear of the implementation of personalized medicine into health care. Individuals with rare disease require personalized medicine now, and it is anticipated that the implementation of new technologies currently used to diagnose and guide care for patients with rare diseases will ultimately be applied to all Canadians to understand the personal aspects of their disease, as well as their response and side effects to therapies.

I want to conclude my remarks with an international perspective. In 2011 the International Rare Diseases Research Consortium, IRDiRC, was launched. This consortium has as its ambitious goals 200 new therapies for rare diseases and a means to diagnose the rarest diseases by the year 2020. Members of this consortium consist of organizations that have committed to invest $10 million or more over five years to research projects and programs related to the international rare disease consortium's objectives. Member groups include the European Commission, several of the U.S. National Institutes of Health, and importantly for us, both Genome Canada and CIHR.

The executive committee is currently chaired by a Canadian, Dr. Paul Lasko, scientific director of the CIHR, as is one of the three scientific committees of this body. IRDiRC held its first meeting in Dublin in April 2013. Canada was well represented with over 15 individuals from CIHR, Genome Canada, Health Canada, CORD, and academic medicine attending.

It's important for us to realize that the rare disease community is a global one, and collaboration will be essential to describe new rare diseases and identify the underlying basis to allow for diagnostic testing, to provide support for patients across borders, and to lead toward development of new, rational, and cost-effective therapies.

Canada is exceedingly well positioned to be a major leader in this field, with the realization that collaboration is critically important in areas of science where historically competition has been the norm.

I thank you for your attention to my initial remarks. I am optimistic for a future where, within Canada's health care system, patients and their families affected by rare diseases will be able to access a timelier and more cost-effective diagnosis, coupled with access to rational and affordable therapies. Our patients deserve this, and ultimately it will be associated with a healthier and more productive population.

Thank you.

The Chair: Thank you, Dr. Innes, for that very clear analysis of rare diseases. We appreciate it and look forward to asking you some questions shortly.

We will now go to our other guest, Ms. Jacquie Micallef. Could you please give your presentation.

Ms. Jacquie Micallef (Manager, Member Relations, Policy & Partnerships, Neurological Health Charities Canada): Thank you for inviting Neurological Health Charities Canada to testify in front of the committee once again. It has been our pleasure to stand before this committee in the past to testify on behalf of the estimated 5.5 million Canadians living with neurological conditions in Canada.

I would also like to take this opportunity to thank the committee for putting forward many of NHCC's recommendations in your report, “Focussing on the Brain: An Examination of Neurological Diseases in Canada”. The support of our recommendations is very encouraging. It gives the individuals and families we represent a renewed sense of hope that the attention that has been given to research in neurological conditions will continue to be supported, and that attention will be equally given to the translation of the new knowledge we're receiving, which will eventually result, in our vision, in a direct impact on the Canadians who we are here to represent.

As many of you who are familiar with our coalition know, we represent 24 organizations that support Canadians with a range of neurological conditions, including neurodevelopmental, neurodegenerative, chronic, and episodic conditions. Among these varied conditions, NHCC also has a strong representation of rare neurological conditions. These conditions are small in number of incidents, but they are immeasurable in terms of impact on the families, individuals, health care systems, and the Canadian economy.

Individuals, families, and organizations impacted or representing a rare condition are often on the losing end of a numbers game. Often, certain numbers of people are needed in order to conduct clinical trials for new drug therapies, health professionals have little experience in practice with their particular conditions, criteria for assessments are often based on the characteristics of more common conditions, and organizations have small funding bases, which lessens their ability to fund research, programs, and education focusing specifically on their condition.

The following are some proposed solutions, ideas, and comments that NHCC would like to put forward on this topic.

The first is the orphan drug framework for Canada. We would like to say that Health Canada’s commitment to developing a framework for the designation, authorization, and monitoring of orphan drugs is a positive step forward in the treatment of rare conditions. NHCC member organizations, especially those dealing with rare conditions, have strong global relationships, in part due to the fact that there's a small number of experts to draw on in any one country, so that makes their international outreach even more critical in finding those champions.

This framework will provide a place to formalize and elevate that level of international sharing, to elevate the discussions on rare conditions, and to bolster pharmaceutical companies’ interest in developing, testing, and marketing new drug therapies for rare conditions. Also, the knowledge translation and exciting innovations in the realm of pharmaceutical treatments will undoubtedly benefit many of those individuals.

Under other treatment options, we would like to mention the following.

On the term “drug” in the orphan drug framework, one comment we do have is that it may not capture the variety of treatment options that are being researched for rare conditions. An example would be retinal eye disease. The expanded innovative treatment options for these types of conditions include the possibility of stem cell therapy, gene therapy, and prosthesis. These possible treatments may not have a pharmaceutical component, and we would see it as very unfortunate if that type of innovation were to be stalled because it wasn’t captured within that framework. We’d like to make sure that either it’s captured or there is a place for that type of innovation to be considered as well.

Communication devices are another exciting type of innovation. Rare conditions that develop in infancy or young childhood—and, as Dr. Innes said, a number of them do—and that limit a person’s ability to speak or write, or to provide common physical movements in order to communicate, can often lead to the person being presumed to have a low intellect, an inability to experience emotion, or a lack of understanding about the world around them, and the assessment tools that are used to determine cognitive functioning often focus on the physical cues.

● (1545)

I'll give one example. We have representation from a group representing Rett syndrome, which is a very rare condition mostly affecting women. It starts in infancy and is usually determined at around six months to 18 months. The daughter of the president of an organization that we have at the NHCC has Rett syndrome and always has been assessed as having the capacity of a six-month-old. She's 25 years old, and they have always presumed that her intellect was not above that of a six-month-old.

But this exciting new development of eye-gaze technology, which allows her to focus her eyes on symbols or words and different pages, has just opened up the world. They were telling me a story about it the other day. She was at a conference on Rett syndrome. She got agitated, was staring at the word “no”, and by using her eyes was flipping through the pages, which is the way this eye-gaze technology works, to say that she didn't want to be at the conference and she did not want to continue to hear about Rett syndrome. Now, a six-month-old would not be able to express themselves in that way.

It's just amazing that with a slight eye movement, these people are able to gain autonomy and to have a better quality of life, and especially so for the caregivers as well, because for someone whose communication is limited, it's a constant guessing game. These families are now saying that this technology is wonderful.

It does cause a lot of fear and anxiety when you realize that some of the care you've been giving is no longer going to work. My contact said, “I've been tucking her sheets around her for 25 years and maybe she doesn't want the sheets tucked in, and now she's going to tell me.” There is that sort of thing, but it's just amazing that she can gain a sense of autonomy and improve her quality of life.

We've seen this eye-gaze technology. There is a clinician in the United States who has been using it with girls and women who have Rett syndrome. Even a child as young as three years old is using this. In one particular study, she indicated that she felt something was wrong, that she was tired, and that she needed to rest her eyes. A 20-year-old woman indicated that she was hungry, even indicating what her food choice was, and that she was thirsty. This technology has been used with over 100 girls and women in this particular study. In concert with appropriate training and augmentative communication services, it has been found that this technology is highly successful.

In terms of our recommendations, we would like to see some investigation into what is available in the provinces in terms of what is covered across the country. We do know there is a cutting-edge technology, this eye-gaze technology, that is now available in Ontario. We haven't done the investigative process, but we need to know what is covered by the provinces, because we do see this as a really important thing for people with this type of condition or any type of communication-limiting condition.

Genetic fairness is another piece of this. Most rare conditions are genetic, as the doctor has pointed out. Genetic fairness is of course central to our discussion on rare diseases. Canada is the only G-8 country that does not have laws to protect its citizens against discrimination based on genetic information, and genetic discrimination impacts on two key areas of a person's life: insurability and employability.

Neurological Health Charities Canada supports Bill S-218, which was introduced in the Senate by Senator Cowan, and believes that this is a comprehensive bill that will provide the necessary protection for Canadians from discrimination based on genetic characteristics. We'd also like to recognize the championing that Libby Davies has done on this issue. We see the bill put forward by Senator Cowan as comprehensive, as I said, and as really covering off that employability and insurability piece.

In your report, “Focussing on the Brain: An Examination of Neurological Diseases in Canada”, this committee has also recommended that the Government of Canada consider using the results of the National Population Health Study of Neurological Conditions in collaboration with the provinces and territories, as the basis of a pan-Canadian strategy for neurological diseases.

The NHCC would like to see the government invest $3 million over the next three years into the NHCC to develop a framework for a pan-Canadian action plan for the brain that would focus on the key areas that we have put forward in the past, such as research, prevention, caregiver supports, and public awareness education, but also for us to be able to expand our base, to look at those priorities, and to reassess in terms of what the needs are of people with neurological conditions in Canada.

● (1550)

It should also be noted that the National Population Health Study of Neurological Conditions also includes a focus on certain rare neurological conditions and that NHCC expects to receive more information on the incidence, prevalence, and impact of these conditions. We believe that evidence will help to support the case for the solutions we've presented today.

The Chair: Thank you very much for your very insightful presentation. We appreciate it.

We'll now go into our round of questions. It's a seven-minute round and we'll begin with Ms. Davies.

Ms. Libby Davies (Vancouver East, NDP): Thank you very much, Chairperson, and thank you to both of our witnesses for being here today.

First, Dr. Innes and Ms. Micallef, thank you for sharing information which I think gives us a very graphic example of how technological innovation can really help someone, such as the young woman you spoke about at the conference, because sometimes it's hard to go from the paper into the practical world and know how this stuff really works.

One of the questions I've had all along is based on the fact that we're hearing about some of these amazing advances in technological innovation. I do think it's an issue and you've touched upon it. I'd also ask Dr. Innes the same question. It's the issue of accessibility, and you spoke about that in terms of testing.

We heard from a witness last week, Durhane Wong-Rieger, president and chief executive officer of the Canadian Organization for Rare Disorders, who spoke from Geneva. She raised as well this issue about the lack of testing that's available for genetic diseases. It's sort of a patchwork across the country.

You say your organization is dealing with that and you're trying to navigate what's going on at the provincial level, what's available, what's covered. My question is, what can we do? We're a federal committee. We're speaking to the federal role in this. The more specific you are in terms of what you think the federal government could be doing would be helpful. I don't know whether it's through Orphanet, because Dr. Innes also touched on it, but what should we be calling on the federal government to do to make sure these innovations are actually accessible?

I don't know how much people have to pay for these innovations. You mentioned that the issue of what's covered and what isn't is a big consideration. How do we grapple with that? Under the Canada Health Act we talk about universality and portability. Sometimes even at the most basic level of getting service in another province you can run into difficulties, but here we're talking of sophisticated level of things that aren't available to people in even one province, depending on where they live. I wonder if you could address that.

Second, on genetic fairness, I do think this is a very key issue related to innovation when we talk about electronic health records, personalized medicine, and where information goes. Could you give us any concrete examples of what you were talking about when you talked about employment and insurability? Could you give us an idea of what some of your members have experienced when they faced discrimination?

● (1555)

The Chair: Ms. Micallef.

Ms. Jacquie Micallef: If I may, I'll start with the second question first, the one about genetic discrimination.

One of the most common examples of genetic discrimination would be Huntington's disease. If your parent has it, there's a 50-50 chance that you will too. Everyone has to fill out an insurance form where you are asked questions about any conditions that are in your family. Huntington's disease would be one that has a red flag attached to it.

Some people feel that if it's in their family they may be denied insurance, or they are pressured in some way, and we have seen this in employment situations as well, to get genetic testing. That's a very personal thing. If a person with Huntington's is found to have the gene, they know at some point during their life they are going to develop this devastating disease. That's a very personal thing. A lot of people don't want to do it. I think it's made people feel that because something is in their family, they're going to be discriminated against, or they are going to be put in a position to receive information that they perhaps are not able to deal with or process. The impact that could have on a family is really unfair. We know that these people are not protected currently.

There was a story of two sisters who wanted to open a physiotherapy clinic, I believe, but Huntington's was in their family and they were denied insurance. They were tested for the gene and they did not have the gene. They went back with this information and then they were accepted.

People shouldn't have to be put in a position of having to receive potentially devastating information like that, and to have to do it because of their livelihood or to fulfill any dream or aspiration that they have been working toward. I would give that as an example for genetic discrimination.

With respect to what the federal government can do, in terms of the technology piece, I think that's a really important question. Unfortunately, sitting here at this moment, I don't have the answer. As I said, I think a lot more investigation needs to be done as to what is being offered.

The other part of the issue is I tried to reach out to a couple of people who are involved in the communication devices piece to ask if they knew what was happening. I had a response from one clinician from the U.S. It's a little bit spotty when it comes to who holds that type of information, and I wasn't able to get it. It's something that NHCC is absolutely committed to looking into further, but I apologize that I don't have anything concrete.

● (1600)

Ms. Libby Davies: Even to say that we need an investigation is helpful. Somebody has to look at that. Can you guys do it alone? I don't know. But it is something we could recommend needs to be looked at. That's helpful in itself.

The Chair: We're very close to your time, Ms. Davies, so we'll go to Dr. Carrie.

Mr. Colin Carrie (Oshawa, CPC): Thank you, Madam Chair.

I want to take this opportunity, Ms. Micallef, to thank you and your colleagues for the input on neurological diseases. I think the study we did was a very encompassing one. Thank you very much for your input.

I want to ask you about one of technological innovations you brought up, this eye-gaze technology. I was wondering what other conditions it could be used for. How early can it be implemented?

Ms. Jacquie Micallef: Are you wondering when a person could begin to use it?

Mr. Colin Carrie: Yes, could a child?

Ms. Jacquie Micallef: The earliest example I've come across, which I included in the brief, is at three years. There isn't a vast amount of information out there on this technology at this point, but three years old was the earliest. There are other types of diseases, dystonia would be another example, where it could be used.

Of course, there are some conditions that it would not be used for. There are types of muscular dystrophy that affect the use of eyelids, and a person's eyes are not able to be open or be gazing in that way, so it would not be effective there. Especially with neurological conditions, some of them.... This is interesting, because it's starting from children. The interesting thing is that when someone develops a disease, an example could be Parkinson's or Alzheimer's, something they develop later in life, we already have an understanding of that person's likes and dislikes, their intellect level, all of that sort of thing. We already have that; that person's sense of autonomy has already been carved out. So when they develop a disease, a caregiver or health care provider, if they're taking the right amount of input and interest in the person, is able to continue care in that way. This eye-gaze technology, especially starting at three years of age, sets the person up to be able to start saying what their likes and dislikes are, and as I said, get that autonomy.

It's important that there be a lot more work done, a lot more studies out there about this information, and to be using it as early as possible.

Mr. Colin Carrie: I found it very exciting that people who you think are “brain-dead” is the term that's used, or something along those lines.... I've heard of it before, and I thank you for bringing that to our attention.

I want to ask Dr. Innes a question as well. You mentioned diagnosis as the first step in managing the condition, and you explained how Orphanet is an international portal supported by CIHR. I was wondering if you could elaborate a little bit. How does the technology, the innovative data collection and the way Orphanet works, help diagnose rare diseases? You talked about this diagnostic odyssey people go through. Perhaps you could tell me how we can start using this innovation of Orphanet, especially with rare diseases, because there's not a lot of information about them to make sure that we get a proper diagnosis.

Could you explain that?

Dr. Allan Micheil Innes: I would certainly view multiple layers or levels to the approach of a diagnosis with rare disease and acknowledge the fact that many physicians, even specialist physicians, will have limited knowledge of several rare diseases. Other physicians, such as myself, who practise in the area of medical genetics may have enhanced knowledge. I think Orphanet, which serves a number of roles, can be an early tool to help with diagnosis. A primary care physician or a family can search Orphanet, recognizing the symptoms that are present in the patient or their child, and can use that to try to guide toward a specific diagnosis. That can obviously help, because not every patient can see a geneticist. That can help set people on the right path. There is certainly searchable information there that can help direct to a specific diagnosis.

Now, many times formalizing that diagnosis may require a visit to a specialist and often confirmatory diagnostic tests, and that's a separate barrier. But we at least see Orphanet as a way to get people on the right track. As a clinician I have the experience of having met many people who are clearly alone, struggling with a lack of diagnosis. I get e-mails from people. I don't even know how they get my e-mail address, but I'm glad that they do. It predates my time in Orphanet. They just find me somehow. They're sitting in their living room somewhere, their child has something, and they've found me, provinces away, and they're asking for my help.

I think families are often isolated, and this is one more tool to help them.

● (1605)

Mr. Colin Carrie: Can you comment on the amount of information that is contained within Orphanet and how, in the last few years...? You mentioned Canada was the second country to sign on. How is it populated, and what kind of data collection do you have? What is the volume of data, just in the last few years, by using this technology?

Dr. Allan Micheil Innes: I'm sorry, but I don't have specific numbers or data points for you. I should clarify that Orphanet, although it had its origins in France, has been fairly ensconced throughout the EU for several years. Although we were the first partner outside Europe, many European countries have been inputting information.

The advantage to that for those of us in Canada who have to join is, among other things, to join Orphanet you have to have all the information available in your national languages. We're already well covered. Also you can imagine that information is there at least in many European languages, and now with Japan and Brazil and others coming on, other languages will be added.

There is a brief summary of essentially every rare disease. It's a moving target, but for the 3,000 to 7,000 known rare diseases, there are at least several paragraphs outlining the basic features of that disease. That's one. It's the encyclopedia of rare diseases. There are also databases for orphan drugs that are available, clinical trials that are going on, and access to registries.

For some of those things you didn't necessarily need Orphanet Canada per se. It's findable on the Internet. What Orphanet Canada has provided for Canadian physicians and Canadian patients is a list of where the clinics are in Canada that have expertise in these diseases, who the Canadian support groups are, the family support groups they can reach out to, what research projects are happening in Canada, and what registries you can participate in. That's the value added since we've joined.

We only launched in October 2012, so we've reached out through our offices to about 350 stakeholders. We're constantly trying to identify more. I think we have data on 70 of those live on our website so we have a long way to go, but we're gathering that information.

The Chair: Thank you.

Now we'll go to Dr. Fry, please.

Hon. Hedy Fry (Vancouver Centre, Lib.): Thank you very much, Madam Chair.

Dr. Innes, you were talking about this way of looking at a broader...almost covering someone's DNA.

How far down is this in terms of research? How far has it gone, and what countries are using it, if at all? If you see it being used extensively, what cost savings do you see being achieved by doing that? That's the first question I wanted to ask.

The second one is, as you well know biologics are being used quite often to deal with some rare diseases. What do you see as the benefit of new drugs coming out that are, for want of a better word, generic versions of biologics that are coming out of certain developing countries? As we look at cheaper ways of dealing with rare diseases, what do you see as the downside to that? I wonder if you see this as a risk that might cause problems because, as you well know, generic drugs don't always have the same molecular structure or even efficacy as some of the pharma drugs.

Dr. Allan Micheil Innes: Thank you for both questions. I certainly can answer the first in more detail and with more comfort based on my level of knowledge. I'll speak briefly to the second. I'm not someone who prescribes biologics personally or uses those drugs.

When we think about personalized medicine, whether it's for rare disease or for the general public, we're eventually realizing that any one given disease, whether it's a rare disease such as Rett syndrome, which has been discussed previously, or the more common diseases, such as breast cancer, rheumatoid arthritis, etc., these will not end up being one disease but many diseases. If a medication is developed with a targeted purpose, targeting that rare disease, then modifying that drug in some way may then be off target. That would be the extent of my knowledge on that matter.

The new technology of next generation sequencing has really been transformative. I would not have predicted its existence even five years ago in my practice, but truly, the opportunity to look at someone's entire genetic code and analyze it now exists. We're using it here in Canada. Canadian researchers have been very successful and internationally competitive in using this technology at a cost that is really close to attainable. It's in the $2,000 to $3,000 range for this type of experiment, which is still a lot of money, but it's not much more than the costs I mentioned earlier of $1,000 to $2,000 to look at a given gene, which is the current way we do it now, one by one, or the cost of an MRI scan or other technologies that we currently use, and these costs are plummeting. I suspect these costs will be less in a few years. It won't be the price that will be the barrier anymore.

There are issues when you sequence someone's entire genetic code to try to find a diagnosis. You will generate enormous amounts of data. You need computers that can handle that and specially trained people who can handle that, and you will identify findings—and I alluded to this briefly in my brief—that are, we could say, incidental. They're not the reason that you did the test. You're examining a child who has a developmental problem and you find that she has a breast cancer gene that might affect her when she's older but might affect her mother now. How do you deal with that?

Then there will be spin-off health care costs to that. You might now be ordering mammograms or breast MRIs you hadn't anticipated. These are not unsolvable problems. The world is thinking about them in a very thoughtful way and Canada is helping to lead that. I anticipate that these technologies will be quite democratized so eventually they will be quite readily available. I think if anything, my plea would be for Canada to lead the world in implementing them in a thoughtful way and not in a non-thoughtful way, because if we do this poorly, we will regret it.

I think we're nearly at the point now where implementing this technology early could save money. It is expensive. Right now you can get these kinds of tests commercially in the States for about $10,000. That's probably too expensive for the health care system to manage, but we're getting close to having it there. When I speak of the diagnostic oddity, the children who I see and the children and the families that the other witnesses speak to often come to the clinic for five or ten years. That's missed work. That's parking at the hospital. That's hospital visits. That's invasive tests, biopsies, MRI scans. The costs often far exceed $10,000, and it's five to ten years of wasted time where we could have had a diagnosis and focused our energy in a different way.

● (1610)

The Chair: You have about one more minute.

Hon. Hedy Fry: Thank you. I have just one more question I want to ask. You probably won't be able to answer it because it's kind of a philosophical question.

What happens if you can sequence the entire genome structure of a human being, of a particular individual, the entire DNA, and you have some of these coincidental findings? Do you think that would lead us to start treating things that may or may not cause a problem later on? Will we begin to start treating people far too early? Will we begin to jump-start everything on something that we think might happen? What are the ethics of that? What is the downside to that? Forget the money. What is the downside?

Dr. Allan Micheil Innes: These are tremendous questions and ones that people are thinking about a lot.

I think there are a few issues and it's hard for me to do it all justice in a brief amount of time. But you're right, in that there are both the pros and the cons. There's clearly the chance that we might identify things that we didn't expect before. We might identify a genetic variant that might predispose to sudden cardiac arrest, where you intervene with a defibrillator and you save a life. On the other hand, you might find a genetic variant, and although that gene is linked to a disease, you don't know whether the variant you found in that patient will cause the disease. You may end up screening them, treating them, medicalizing them for conditions they may never develop.

There are lots of ethical issues about genetic testing in children or in adults for many of the reasons that the previous questions have highlighted, including insurance. We feel quite strongly as a community that we shouldn't be testing children just to give them information about what genes they might carry that would affect their offspring, for example, or to identify a gene that might tell them that they're going to develop breast cancer when they're 60, or Alzheimer's when they're 80. That's an adult's right to make that decision, not a child's.

We have to be thoughtful about those things.

● (1615)

The Chair: Thank you so much, Dr. Innes. You made some very good points.

Mr. Brown.

Mr. Patrick Brown (Barrie, CPC): Thank you, Madam Chair.

Thank you to the witnesses for the testimony today.

It's nice to have Neurological Health Charities before us. I was on the committee, along with Ms. Smith, when we had the subcommittee on neurological disorders. I know that Shannon from your office was very helpful on some of the input that your organization put forward. It's great to have your input on this study on technological innovation.

I want to get your perspectives on how Canada is doing when it comes to international collaboration. I know we have a lot of hope for some of the work that's being done abroad with Germany and France and the U.K. on having that broader population study.

Are we doing enough of that type of collaboration that enables research and innovation?

The Chair: Go ahead.

Ms. Jacquie Micallef: Sorry, I'm just thinking about my answer.

The Chair: Oh, here you don't dare think. You just have to talk.

Voices: Oh, oh!

Ms. Jacquie Micallef: You just speak.

The Chair: I'm just teasing.

Mr. Patrick Brown: I guess what I mean is that I see we're doing this on Alzheimer's and dementia, but are there some broader trends we can be looking at, or are those the types of partnerships that you believe work?

Ms. Jacquie Micallef: You're right in that we're seeing a lot of condition-specific global partnerships. I think with the development of NHCC, and with these various neurological conditions coming together, it's made us realize, even more so, and now that we're about four years in, that the commonalities between these conditions are so similar.

Now we are getting the results from the study and we're starting to move into the synthesis. I think over the next couple of months we'll have a better idea of, obviously, what the findings are, what some of the gaps are, and what some of the opportunities are for research further down the road.

I think it's absolutely our aspiration that we would keep it at a level of the commonality between the neurological diseases, at the same time recognizing that condition-specific is absolutely needed.

Mr. Patrick Brown: I'd also be interested in your perspective on this aspect. When we talk about CIHR, which obviously is the wing for research that your organizations and your members do a lot of work with, and when it comes to technological innovation, do you think enough of CIHR's resources are put towards technological innovation?

What type of balance would you recommend in terms of direct research and clinical trials and those more tailored towards technological innovation, if you look at new devices and new means of doing things?

Ms. Jacquie Micallef: That's a very good question, and I don't think I have the knowledge to answer it. I apologize.

I'd be more than happy to get an answer to that question for you.

Mr. Patrick Brown: No, no, I appreciate that.

There's a question I want to pose to both Jacquie and Allan. It's a question I've asked panels before. We've had a series of panels on technological innovation.

When it comes to areas that are strictly in the federal jurisdiction —a lot of health care, obviously, is provincially administered—one area that is a tool for innovation is the regulation of medical devices. One doctor who came in here said it was a slow process, and another said it was excellent compared with the U.S., where he had dealt with this.

What are your perspectives on the regulation of medical devices with any of the organizations or groups that you've worked with? Are there improvements that can be made in Canada, or do we have the right balance right now?

Ms. Jacquie Micallef: I'll let Dr. Innes answer that.

The Chair: Dr. Innes.

Dr. Allan Micheil Innes: Thanks.

I also feel that I perhaps am not qualified to answer that question. I'm not one who's worked with a lot of groups in implementing devices.

Diagnostic tests are a slightly different matter. I'm actually not aware of Health Canada's jurisdiction in that area, but I think we would certainly make the plea, when it comes to diagnostic tests, that these are medical devices.

I make the point that “recreationally”, if I can use that word, you can get some of your genetics tested if you swab your cheek or spit into a saliva kit and send it to some companies in the U.S. They will ostensibly give you medical information back, which is worrisome to us. They've repurposed those as being basically recreational results in letting you know what your eye colour is and whether you can curl your tongue and such, but also buried in there is actionable medical information which I think should be administered through the appropriate channels.

It's those types of things. I think medical diagnostic tests should always be recognized as what they are.

● (1620)

Mr. Patrick Brown: Jacquie, I have a final question for the members of the Neurological Health Charities.

We can learn a lot from other countries in terms of there not being a monopoly on a good idea. Just as other countries pick up ideas in Canada, do you, whether it's the ALS, Parkinson, or Alzheimer societies, have ongoing dialogue with associations in different countries? Do you know of any new technological advancements that might be happening elsewhere in the neurological field that we should be aware of in Canada or that we should invest in ?

Ms. Jacquie Micallef: That is something we're starting to look into more now that we are coming to a point where we're wrapping up the study and we're beginning to look at what the next steps are and broaden our horizons.

Before joining Neurological Health Charities Canada I was with the Alzheimer Society for six years. I just joined the NHCC in January, so it's still very fresh. There is an international Alzheimer body, as there is for many of the other diseases. The international congresses are happening. A lot of them are actually happening now, over the summer and into the fall. So I think that dialogue is happening.

I will take that back, because I know from my experience, we've looked a lot at service delivery in other countries, and it's very difficult to try to take service delivery in another country that's emerged in a very different way, is a very different culture, and place it here. But in terms of this topic, in terms of technology and devices, I think there is a lot that we could learn. As I said, in moving forward and expanding our horizons, it will absolutely be something that we take forward.

Mr. Patrick Brown: Thank you.

The Chair: Thank you so much for your insightful answers.

Mr. Brown, it's really nice to hear you on those questions on neurological diseases. Mr. Brown was a great asset on that committee.

We'll now go into our five-minute rounds, and we'll begin with Dr. Sellah.

[*Translation*]

Mrs. Djaouida Sellah (Saint-Bruno—Saint-Hubert, NDP): Thank you, Madam Chair.

My thanks to our guests.

I know there are a lot of rare diseases, as many as 3,500, and more than 80% of them are linked to genetic factors.

I will get right to the point. In Quebec, we have a newborn screening program, which seeks to detect conditions such as phenylketonuria, congenital hypothyroidism or a deficit of acyl-CoA dehydrogenase in medium-chain fatty acids in the 24 to 48 hours after birth.

Could you tell me if similar tests are done anywhere else in Canada? If so, that’s great.

Are there also other tests being systematically administered at birth?

My question is for Dr. Innes.

[*English*]

The Chair: Dr. Innes.

Dr. Allan Micheil Innes: Certainly, newborn screening is one of the major public health successes of the last 50 years. I would say that every province in Canada, indeed almost every jurisdiction that I'm aware of, at least in the developed world, does have some sort of newborn screening in place.

The panels differ from province to province, and those are under the purview of the provinces to decide, although there certainly are some common conditions, including phenylketonuria or congenital hypothyroidism, which are essentially screened for universally. The purpose of these programs is primarily to screen for conditions that are eminently treatable in the neonatal period, for which the children have a pre-symptomatic window where they're completely well, but if exposed to either a certain toxin in their diet—it can be things that are well tolerated by normal children—or in the absence of a hormone such as thyroxine, their development will regress in an irreversible way.

That's the principle of newborn screening, which is universal across Canada, to identify a small subset of diseases for which there's a rapid, usually a relatively cheap, therapeutic intervention. Where there becomes a little bit more tension is on whether you expand that screening to detect also conditions that are untreatable. There are arguments pro and against that.

● (1625)

[*Translation*]

Mrs. Djaouida Sellah: According to Orphanet, people with rare diseases are more psychologically, socially, financially and even culturally vulnerable, particularly because their challenges have to do with being able to access quality healthcare, comprehensive social and medical support, effective liaison between the hospital and general practice, as well as social and professional integration and autonomy.

Innovative technologies could be particularly beneficial for people with rare diseases, but the rarity of the disease can create strategic difficulties. In your view, how should we balance the high costs of research and treatment of low-impact and low-prevalence diseases and the great need experienced by this small patient population.

[*English*]

The Chair: Who would like to comment on that?

Dr. Innes.

Dr. Allan Micheil Innes: Thank you for that question.

Obviously, I speak both as a coordinator for Orphanet Canada and a researcher but also as a physician for individuals affected by these rare diseases. As their physician, it's nearly impossible at times to resist the urge to embark on further research to get answers for what are truly devastating disorders. Having said that, we do need to recognize at times there's a limited pool of resources, whether that's research or clinical dollars.

I think a few points can be made. One that I already alluded to previously is that occasionally research into a rare disease can often give us tremendous insight into common disease. This story has played itself out many times over in the fields of hypercholestor-alemia, and many rare cancers. Through the research into rare genetic forms of cancer, you find the genes that are also the same genes that are mutated in people with common cancer. One point is that the research is often generalizable.

I think going forward, if we're talking about 3,500 rare diseases, the system will be stretched if we're talking about 3,500 new expensive therapies. What I think we may have to envision is collapsing down various rare diseases into families or groups of diseases where maybe you can have a common therapeutic approach.You may have to pool things, given the limited resources.

The Chair: Thank you, Dr. Innes.

We'll now go to Mr. Wilks.

Mr. David Wilks (Kootenay—Columbia, CPC): Thank you, Chair, and thank you to the witnesses for being here today.

Dr. Innes, you mentioned next generation sequencing in your presentation and you mentioned 180 pediatric disorders that have been identified. Where do you see next generation sequencing going in the next five to ten years? What do you think the federal government can do to work with that technology?

Dr. Allan Micheil Innes: Next generation sequencing certainly has a variety of applications that are fairly exciting right now. They're right on the cusp of research and clinical. One is the type of thing I've already spoken about, which is for both research and diagnostics into rare disease. There's no question it works there.

The other where I'm a little less personally experienced but it has probably been the biggest success story to date is in cancer. Again, if we realize that all cancer is a genetic disease in the sense that it's a mutation in the gene in that cell that causes the disease, by doing next generation sequencing on tumours, you can identify the genetic changes that are unique to that tumour, and target therapy. That's not my area of expertise, but that's currently in clinical translation.

I suppose the next step is will every patient, will every Canadian, have their genome sequenced at some point, and at what stage in the life cycle? I'm not sure. As a geneticist, I'm not pushing for that yet, but that could come.

I think where we would ask for thought nationally.... It may not become a funding issue eventually. These are still expensive technologies now. There's going to be investment in infrastructures required. The cost will come down, but I think there needs to be guidelines in place for how to implement this technology. We talked already about genetic insurability and discrimination. This can only get worse. You could potentially imagine that if everyone has their genome sequenced, we're all going to be found to carry something. Eventually, the insurance companies will need to realize that. All their clients carry something, but there may be a window or time when things get worse, not better. I think this speaks to that need again.

This is outside the purview of high tech, I suppose, but I would also put in a plea for education. We're going to require a whole new generation of young Canadians who are savvy in computers, bioinformatics, genetics, genetic testing. That's going to require an investment as well.

● (1630)

Mr. David Wilks: I will give the rest of my time to Mrs. Block.

The Chair: Okay.

Mrs. Kelly Block (Saskatoon—Rosetown—Biggar, CPC): How much time is that, Madam Chair?

The Chair: You have about a minute and a half.

Mrs. Kelly Block: I'll be quick.

I had the opportunity to attend a fundraiser, probably a year ago now, for a young girl in my riding who has Rett syndrome. I understand that about 17 girls in Saskatchewan have this rare disorder.

It's just overwhelming when we hear that we are looking at over 3,500 rare diseases or disorders. You spoke earlier in response to a question from my colleague about collapsing the therapy, about how you would manage some of these rare disorders by grouping them into families. I wanted to let you expand on that a little, Dr. Innes.

Dr. Allan Micheil Innes: Thank you.

I guess we could think of it in two ways.

My colleague has already spoken very nicely about non-genetic technologies that can be used, so if these new technologies can be leveraged regardless of the genetic cause.... But if we're thinking about understanding why the gene goes wrong and causes a disease and how we can fix that, we have to recognize that sometimes genes work in pathways, so there may be a whole sequelae of genes. You can modulate one or the other, but they may eventually all converge on a pathway somewhere down lower. If you can modulate that target, you may have a successful treatment of 10 or 20 diseases along the pathway. I think we're going to have to think in that way. Researchers may not be able to work in their silo just studying their one disease. They may have to think about a category.

Mrs. Kelly Block: Thank you.

The Chair: Thank you, Mrs. Block.

We'll now go to Mr. Kellway.

Mr. Matthew Kellway (Beaches—East York, NDP): Thank you, Madam Chair.

I thank our guests for coming today.

Dr. Innes, I was curious about your comments when you said that not everybody can see a geneticist, and the kind of random discovery of you by some people, somehow.... As the technology develops, and you touched on this a bit in response to Mr. Wilks' question, I'm wondering if you have any thoughts about how this plays out for people who have a child or who themselves develop certain symptoms. How do you get to a geneticist to take advantage of the diagnosis and potentially therapies and treatments?

Dr. Allan Micheil Innes: My point of course is that everyone is welcome to see a geneticist if they meet criteria, but our numbers are limited and the waits are long. Also, as you say, even many physicians, let alone families, don't know of our existence.

Data portals like Orphanet can help. Tools to get an earlier diagnosis can help. We can leverage technologies; we're already using them. Our clinics are very involved in outreach. I travel throughout the province to see patients. We use telehealth when it makes sense. There are ways to bring ourselves to the patient. There may be ways through more democratic availability of sequencing or using computer algorithms to get diagnoses more quickly.

You may never be able to replace the geneticist or genetics counsellor as being the doctor or the person with the most expertise in that disorder, who still needs to talk to you about it and walk you through the natural history. That may always be a bottleneck, unless we can train more of us, but there's a variety of ways in which we can get information out to people so that they can at least get to us, or so we can start working with them to get the information we need, so that when we do see them it's not a wasted visit.

● (1635)

Mr. Matthew Kellway: Is there discussion in the community about ensuring that there is a growing number of people getting trained in genetics? I mean, if this is the future of medical science, is somebody thinking about growing that profession or expertise? Do you know?

Dr. Allan Micheil Innes: Another hat I wear is that I chair the specialty committee for medical genetics at the Royal College of Physicians and Surgeons of Canada. It is part of our purview, along with our specialty society, the Canadian College of Medical Geneticists, to think about human resource planning, both for medical geneticists and for genetic counsellors.

It's a complex issue in regard to how you train a physician, which takes a long time, and then ensure that physicians have a place to be hired to. It may seem paradoxical, but it is a fear for these trainees when they're done, unless there's a job.

We're working on those issues, to be sure, but I think that even if we doubled or tripled the number of geneticists, if we're thinking about getting genetic testing out to all Canadians, there will never be enough. My profession is going to have to take a more active role in educating all physicians about what this technology means and how to use it. We're also thinking about ways to do that.

Mr. Matthew Kellway: Thank you.

Ms. Micallef, genetic science seemed to approach so rapidly and changes so rapidly. The bill you were talking about on genetic information and discrimination based on genetic characteristics, how far down the road do you think that's going to be able to work and handle changes or rapid advancement of the science?

Ms. Jacquie Micallef: That's a very good question.

The way we see it, it's about privacy, and it's about protection. Because we have identified those two major issues, being employability and insurability, and to my knowledge we haven't seen it play out in other major aspects of a person's life, I think it's covered there.

The other piece is that putting in a bill like this, with genetic fairness or anti-genetic discrimination, legislation will also help to bolster Canadians' involvement in research. Really, it's sort of flipping it, because it will actually help in some way possibly to move the science down the road. It's a big barrier for a lot of people to participate in research in Canada.

The Chair: Thank you very much, Ms. Micallef.

Now we'll go to Mr. Lobb.

Mr. Ben Lobb (Huron—Bruce, CPC): Thank you very much.

Thank you to our guests for being here today.

I have one question. I'm trying to figure out how to put this best. I was thinking back to a couple of years ago when they did some genetic sequencing for Ozzy Osbourne. You may remember that. It was in the news a couple of years ago. They were doing some sequencing and so forth to try to figure out what made this guy tick and how he's still on earth.

At the time, though, I remember they mentioned the cost, but it seemed to me it was in the hundreds of thousands of dollars. Is that right, or was it in the tens of thousands of dollars to have it done privately?

Dr. Allan Micheil Innes: I didn't expect an Ozzy Osbourne question today, but let me take it on.

You're right. I believe that was maybe three or four years ago and the costs would have been in the $100,000 range. Three or four years before that it would have been $1 million.

The human genome project to sequence the first genetic code of the first person took 10 years and cost $5 billion. We've gone from $5 billion to $1,000 in about 15 years. The pace is quite extraordinary and the costs are plummeting in that sort of scale.

Mr. Ben Lobb: With a diagnostic test like that, what can we find out, basically? What length of details can we find out with that? How much detail can we find out from a test like that?

Dr. Allan Micheil Innes: I think the test performs best in conditions that are clearly genetic. I think that's where it has its highest role right now, so it's an excellent diagnostic test for many of the conditions my colleague and others have spoken to. For highly penetrant genetic conditions that affect children or adults, disorders that clearly run in families, disorders with a strong genetic predisposition, it's very good.

There is clearly a genetic basis to almost every common disease, which is not to downplay the role of environment that is, of course, significant. Our chance of developing diabetes, cancers, heart disease, etc., are at least in part predicted by our genetics. It's my personal opinion, not shared by all, that yes, we can sequence and find out what some of those variants are right now, but we don't really understand how they all interact with one another to create a risk for an individual. I'm a bit more guarded about that outcome.

We may get to that point, but I don't see that as the usefulness right now. It's more about the risks of strongly genetic conditions.

● (1640)

Mr. Ben Lobb: Forgive me if you've already mentioned this, but is this currently being done in Canada on a proactive basis, or is it not?

Dr. Allan Micheil Innes: Not clinically, although we are starting to pilot that. I didn't have the opportunity to mention that we do have a large Genome Canada and CIHR funded grant on rare diseases. We're going to start piloting it. I believe there are other jurisdictions, including the SickKids Hospital, where they're looking at that, but it's not on the ground clinically in a widespread way in Canada yet. But it will come.

Mr. Ben Lobb: There's an ethical part about it, I guess, between being a doctor and everything else, and trying to play God. At the same time, there must be quite a sound case financially to perform some of these tests proactively, whether they're on identified at-risk children, to try to get them the help they need, or at least monitor them before their condition flares up or shows itself. Is that the discussion they're talking about at SickKids?

Dr. Allan Micheil Innes: I think they're thinking about the same sorts of things that we're picturing. You're right in that there are ethical issues that can't be trivialized, and I'll try to reassure you that the community is thoughtful about them.

Right now we're primarily talking about seeing children who are presenting with something that's wrong with them and we don't know what it is. In many cases, we don't know what the future is going to hold for them. The idea is that if you can identify the answer more quickly, you can get them on the right path. That may not be a cure, and it may not be a treatment, in the sense of a fancy device. It may be that the child needs an ultrasound or something other children don't need, or perhaps new interventions in the school.

If you do those tests for diagnosis, you may also stumble across other things that weren't the main reason for doing the test but are also relevant for their health. We feel we are obliged to deal with those if they affect their health in childhood.

If we scale it out to the bigger population, you could see at some point asking for this for all children who are healthy before they get sick. We're not there yet, but we need to think about it so we don't do it badly or rush into that.

Mr. Ben Lobb: In the United States right now—

The Chair: I'm sorry, Mr. Lobb. Your time is up. You asked good questions, but we're a little over time.

Thank you. Sorry about that.

Mr. Chisholm, you're next.

Mr. Robert Chisholm (Dartmouth—Cole Harbour, NDP): Thank you very much, Madam Chair.

Ms. Micallef, you spoke about the different levels of coverage in the provinces. When I think of health care in this country I think of the real discrepancies that exist from one province to the next in terms of the level of services that are available, as well as the coverage.

I wonder if you could give us some indication, perhaps from both organization's perspective, of what you're doing to help a family who has a child with a rare disease access treatment, resources, and supports, in the ways that you both talked about in your presentations.

Do you help coordinate support? Do you help communicate? You talked a bit in your presentations about how you have various registries set up, and so on, in order to do that.

How do your organizations help these families deal with a family member who has an identified rare disease?

● (1645)

Ms. Jacquie Micallef: I'm from Neurological Health Charities Canada, so I'm representing a number of organizations. Within our organizations, that type of direct service work absolutely happens.

It's not a rare condition, but an example I could give is the Alzheimer Society. If you're an adult diagnosed with any type of disease or rare disease, especially of a neurological type, all of a sudden your job is in jeopardy. Your ability to provide yourself with housing and your driver's licence are at risk. Transportation is a problem. It's not like being diagnosed with the chicken pox or a broken leg. It is going to be with you for the remainder of your life. A lot of the cases are degenerative, and it affects your life in a major way.

One thing the Alzheimer Society has is a program called first link. It's across Ontario, and now they're trying to expand it nationally. It's a connection between the organization and the diagnosing physician in most cases. Upon diagnosis, the family gives consent for their information to be sent to their local Alzheimer Society, and that's so it doesn't get to a crisis point. The person isn't left scrambling, trying to catch up with what's happening as the disease progresses. You get that support right away.

We see a lot of caregivers burning out. People are being dropped off in emergency rooms. Then we have this huge issue of people being admitted into hospitals and staying in hospitals when they don't need to be.

I would give that as one example.

In terms of the rare conditions, it's really difficult. The organizations are small and the funding base is small. These charities run mostly on donor dollars, and people donate because they're impacted personally by a condition.

As I said, it's a numbers game. When you don't have that many people who know about the disease, it becomes really difficult to get those types of services. It's often up to families, like the example I gave with Rett syndrome. The president of Rett Ontario is the mother of a 25-year-old daughter. She is trying to figure out who has it in her own community, how she can help them, and what they can learn from her experience. It's much more organic.

I'll give some time to Dr. Innes here.

The Chair: Dr. Innes, would you like to make a comment?

Dr. Allan Micheil Innes: Thank you.

Orphanet Canada is not resourced, per se, to help those families with the day-to-day activities. In that role, all Orphanet Canada can do is help direct them to the relevant organizations. But this is something I deal with in my practice.

Rett syndrome has been mentioned a few times. I heard the number of 17 young women in Saskatchewan. That's a rare disease, but that's still 17 people who can have a focus and a mission.

I see individuals with conditions where there might be 17 people in the world who have it or fewer, or their condition doesn't have a name so there is no group to coalesce around them, and you have to somehow still champion those people. That's really difficult, especially working in an overburdened health care system.

I don't have the final answer, but there are all these pockets of rare islands of people who do need help.

The Chair: Thank you, Mr. Chisholm. You've contributed greatly to our committee.

Mr. Lizon.

Mr. Wladyslaw Lizon (Mississauga East—Cooksville, CPC): Thank you very much, Madam Chair.

Thank you, witnesses.

The first question I have is for Dr. Allan Innes.

Could you tell us how Orphanet works? I know that it's a portal. It looks like it's more of an informational portal. It exists in many countries. How do you collaborate? What information goes on? How often does new information go on about what's relevant, what's not relevant, and who decides what is relevant and what is useful for people?

Dr. Allan Micheil Innes: Absolutely, Orphanet is primarily an information portal. That's its major purpose.

There is both national-specific content and international or global content. The international global content is out of my control. That's not to say that I or any other interested Canadian couldn't advise them and say that some disease information looks out of date, or to add this disease or that disease, but that's well organized at a higher level and they constantly curate that information on rare diseases.

For Orphanet Canada, we can do a few things. One, we use the site to advertise activities around rare disease. That can be presentations, café scientifiques, research projects, and announcements. When the minister made an announcement about orphan drugs, we viewed that as a way to get that information out to people.

Also, one of my main jobs as national coordinator is to curate information about the support groups and the clinics that exist. I have a small nucleus, my scientific advisory committee. We need to make sure that information is accurate. It's not enough for a doctor to say, “I'm an expert in that condition.” It's not my job to comment necessarily on the competencies of every physician, but we need to at least make sure that this is true, that this is a safe study, and this is safe information. We look at that and we want to get as many organizations out there as possible, but it needs to be safe.

Basically we're looking at constantly curating that information to make sure it's up to date about what research, what projects, and what registries are happening in Canada.

● (1650)

Mr. Wladyslaw Lizon: Thank you.

In your presentation you mentioned personalized medicine. My second question is, could you maybe expand on that topic? What is it exactly? Is it based on a genetic code? What is it based on? How expensive is it?

Dr. Allan Micheil Innes: Those are excellent questions.

I think personalized medicine means different things to different people. I don't think we can attach a cost to it, per se, although the proponents of it would say that in the end it's surely more cost-efficient.

It's the recognition that if you take even a common disease, like breast cancer, or anything for example, for years the way we had to do it was to develop a clinical trial. We would try a drug on 3,000 women with breast cancer. Some would respond to the drug. For some there might be deadly side effects to the drug. Many people might have been cured by that drug, but because a certain percentage had a bad side effect, the drug was not viewed to be safe.

Is there a way to recognize which of the people are going to respond to that drug because either their genetic code is different or their tumour is different, and which people are going to have a bad side effect to the drug because they have a genetic enzyme or something that doesn't break that drug down?

This is the sort of thinking, that any disease is not one disease, or a disease occurs in the context of a person. You can build other things into personalized medicine—their society, their home, whatever you want it to mean—but it's a recognition that disease is not one big uniform category, but often a subset of diseases, and every patient has to be viewed differently.

It's a change of thinking, so there will be costs to implementing it. Surely a lot of people will think in the end that costs will be saved because we'll be putting people on the right medication and on the right regime at the beginning and not at the end.

This is happening a little bit right now. For common blood thinners prescribed in the hospital, like warfarin, which people often have to take after a blood clot, it's recognized that people break this down in different ways based on their genetics. You can prescribe the drug differently right from the get-go based on some simple genetic factors. So there are some early examples, but we have a long way to go before this is really ensconced in care.

The Chair: Thank you very much.

Now we'll go to Ms. Block.

Mrs. Kelly Block: Thank you very much, Madam Chair.

I want to thank both our witnesses for being here today.

I appreciate that it could probably be fairly frustrating when you take a look at the scope and magnitude of the work that needs to be done in rare diseases and neurological disorders.

I want to get back to some questions with regard to Orphanet. I have a bit of an understanding now that Orphanet is a repository of information where people, and I don't want to say they dump information into this portal, but it comes in internationally as well as nationally. You said you are the curator for the national information that comes in, or for the national repository. Do you partner with other organizations, like CIHI, or other people who are gathering information, to figure out how you can coordinate what you're doing? I know you said that your aim is to help improve the diagnosis and care and treatment of patients with rare diseases. Other than gathering the information, how do you do that?

Dr. Allan Micheil Innes: These are fair questions. I think each organization has to do what they think they can do to help.

Given our current funding and mandate and time, it's not Orphanet's job per se, or mandate, to itemize the number of patients with this disease or that disease, but to facilitate the existence of those registries and to connect the patients with the stakeholders. We are not in a position to maintain registries for 7,000 rare diseases.

I think what we need to think about for those things, and I talked briefly about the International Rare Diseases Research Consortium, IRDiRC, of which Canada is a major partner.... It's a little bit a side of Orphanet, although there are clearly places where these things interdigitate. They had their first meeting in Dublin, which I attended. I'm part of the working group on registries. I think we need global registries on rare diseases, but probably those registries don't contain very much information, the basics: name, gender, a way to identify those people, make sure the diagnosis is correct, so if there are new treatment trials, those people are findable. Then I think it may be up to local groups, local charities, local governments, or provinces to keep whatever deeper data they're able to curate and manage.

Maintaining databases is pretty labour intensive, and I'm not an expert in it, but unless you're able to maintain and constantly update it, you maybe shouldn't get into that business.

● (1655)

Mrs. Kelly Block: Thank you.

The Chair: Thank you, Ms. Block.

Dr. Fry, you're our last questioner.

Hon. Hedy Fry: Thank you very much, Madam Chair.

I'm going to go back to something that's been niggling at the back of my brain. It's an ethical question. Of course, as a physician you know the *non nocere* thing, and you don't want to harm anyone. Obviously, if you can help people who have rare diseases, if you can understand what causes the diseases, if you can find treatments for them, if you can put people in touch with others and they can form support groups, etc., this is all excellent.

Given that we can now get somebody's complete DNA profile, is any work being done whereby one can do this in utero through amniocentesis, through some way of taking cord blood, or something from the baby in utero, to look at this? If one is able to say that a child is going to be born with a rare disease, let us imagine the situation where, because rare diseases are going to cost so much to treat or to maintain that person with a healthy lifestyle throughout their life....

Do you remember when people used to say that children who were born with Down's syndrome weren't compatible with life? I'm talking about many years ago. Now we know that Down's syndrome kids grow up and live really happy adult lives. Do you see there being this negative thing where people will start saying maybe we shouldn't...maybe we should consult with the mother to abort this baby. Or maybe if someone is coming up with some DNA profile, the coincidental stuff that we find out turns out to be somebody with a gene that says they're going to be an axe murderer or serial killer. Do you see this kind of thing leading to those negative outcomes?

What are you doing as a group to look at the ethics of this and to find a way to take the benefit from this, while guarding very strongly against *non nocere*?

Dr. Allan Micheil Innes: Thank you. It's a tough question. It's a question that is hard to answer in a few minutes. It's a question that hits close to many of us and what we do already, in the sense that we are faced with these ethical issues that are not easy.

I will say that in many cases I'm remarkably impressed by the ability of parents to make thoughtful decisions about their children. I think what is often assumed is that if the technology exists, families will always go in a certain direction, and that's not true. Parents very much love their children and they're—

Hon. Hedy Fry: I think families will make their own personal decisions. But I'm wondering, could the state suddenly say "My gosh, this person is going to be a serious serial murderer and we don't want this child...? I mean, there are things where states can do that. We've seen that happen.

Dr. Allan Micheil Innes: Well, yeah.... The history is there in many jurisdictions, including my own province, so you're right. I guess I would have to reflect back and look at my colleagues who are in government and hope that's not the case. I guess it's a theoretical fear. I don't see that coming. I think these are always going to be decisions between parents and their doctors.

I will add that this technology will advance, and the ability to test pregnancies in different ways will come, and it will come in the U.S. Again, I think it's simply a matter we should think about at a legislative level and a thoughtful level.

● (1700)

The Chair: Dr. Innes, thank you very much.

That was, indeed, a very insightful question and an important one.

I want to thank Ms. Micallef and Dr. Innes for coming today and contributing very much to our technological innovation study.

I want to thank the committee members as well. I understand the bells are going to ring at 5:15 p.m.

The meeting is adjourned.