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Chair

Mr. Bill Casey

Standing Committee on Health

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● (0830)

[*English*]

The Chair (Mr. Bill Casey (Cumberland—Colchester, Lib.)): Welcome, everybody, to the 118th meeting of the Standing Committee on Health.

Today we're going to continue our study on rare diseases and disorders. We have four groups to make opening statements. We welcome you all.

Today, speaking as individuals, we have Dr. Michael Brudno, Professor and Scientific Director, Centre for Computational Medicine, Hospital for Sick Children; together with Ian Stedman, Osgoode Hall Law School, York University. From the Children's Hospital of Eastern Ontario, we have Dr. Alex MacKenzie, Clinician Scientist. By video conference, speaking as an individual, we have from Toronto Dr. Joel Lexchin, Professor Emeritus, School of Health Policy and Management, York University. From Janssen Inc., a Pharmaceutical Company of Johnson and Johnson, we have Stacey Silverberg, Stakeholder Engagement Manager, Government Affairs and Market Access; and Jacqueline Dobson, Government Affairs and Policy Manager, Government Affairs and Market Access.

We'll open in that order and ask Dr. Brudno to begin.

Dr. Michael Brudno (Professor and Scientific Director, Centre for Computational Medicine, Hospital for Sick Children, As an Individual): Good morning, and thank you for the opportunity to speak to you today about genetic testing and its role in enabling access to therapies for Canadians affected by rare disease.

I'm a professor at the University of Toronto's Department of Computer Science and the director of the Centre for Computational Medicine at the Hospital for Sick Children. My team and I work to develop computer software and algorithms to assist rare disease clinicians and patients, and we have been involved in many Canadian and international rare disease genomics efforts.

With me today is Ian Stedman. He's a rare disease patient and parent who went through a 32-year-long diagnostic odyssey to get an answer for him and for his family, and he will talk about the cost of being undiagnosed and the value that the diagnosis brought to him.

To begin, I would like to talk about the importance of genetic diagnosis for rare diseases. Our genome contains 20,000 genes, and 80% of rare diseases are caused by changes or mutations in one or a few of these. We now have technology that can decipher the whole genome, identify disease-causing mutations and differentiate between the greater than 7,000 diseases for as little as $1,000.

As you heard from Marc LePage, the President and CEO of Genome Canada, on October 16, countries around the world are embracing this technology. The U.K. government plans to decode the genomes of five million patients as part of a national initiative that builds upon the completed “100,000 Genomes” study. The United States just announced a $1.5-billion program through the NIH called “All of Us”, which will combine the genomes of one million Americans with data on their health, environment and lifestyle to identify optimal treatments.

These programs exist because precision medicine will lead to significant cost savings through early intervention and treatment. This is especially true for rare diseases, where patients undergo a significant diagnostic odyssey. Recent studies demonstrate that early diagnosis of rare disease patients through genome-wide testing leads to cost savings of as much as $8,000 per patient per year, in a health care system very similar to Canada's. The reason for these savings is that patients without a diagnosis do not save the health system money by going untreated. They cost the system money because they are the cause of unnecessary visits, investigations and interventions.

Ian can illustrate this with his story.

Mr. Ian Stedman (Osgoode Hall Law School, York University, As an Individual): Good morning and thank you.

As Professor Brudno said, I went 32 years without a diagnosis. Throughout the time I was sick, I had arthritis, headaches, full body rash, fevers and a generally low sense of self-worth, to be honest. I went to my doctor's office—my family doctor—about 200 times before I was 18 years old. I went to see over 30 specialists, had countless ER visits and tons of tests. The answer was always, “We don't know. Try these drugs and we'll see if we can treat your symptoms and let you live.”

Everything changed in 2012 when my daughter was born, because she was born sick as well. We ended up at SickKids and we got genetic testing. It turns out that we have a one-in-a-million disease called Muckle-Wells syndrome. It's caused by a single gene mutation.

That allowed us to get access to medicine, and the medicine, like a light switch, turned off all the symptoms overnight. There has been no looking back—32 years of struggle disappeared like it never happened—so I get to live the rest of my life disease-free. My daughter was only one then. She's six now. The only way she knows she's sick is that a nurse comes to our house every eight weeks and gives her a needle. Otherwise, it's smooth sailing.

Diagnostics, genetics, it's amazing, life changing. It's sad to think that we're the lucky ones now. Hopefully, that isn't the case going forward.

Thank you.

Dr. Michael Brudno: Ian is a great example of why genetic testing is so important and why we need to enable access to genetic testing.

Since receiving treatment, Ian has become one of Canada's scholars in the area of parliamentary ethics law and has published extensively on the topic. He also holds a fellowship on artificial intelligence, ethics and law and is researching how artificial intelligence solutions can be implemented and regulated in our society. I would submit that the cost of his testing and treatment pales in comparison to his contributions to Canadian society. He and countless patients like him are why genetic testing is so important, and why it's important to help eliminate barriers to access to treatment.

Genetic diagnosis can open the path towards therapy, as it did for Ian; however, the diagnosis must be timely, as one can miss the optimal treatment window. Ian, for example, had a one-third chance of dying from amyloidosis by the time he was 35 if he did not receive treatment.

In many other diseases, the window for treatment is even smaller. As a result, many hospitals in the U.S. are now experimenting with rapid genomic testing, with a result within 48 hours for patients in neonatal intensive care units. This also has been shown to actually reduce overall costs because of a reduction in the myriad of tests that would otherwise follow.

I want to now turn to the issue of the costs of these tests and drugs. While the sticker shock of the price of genetic testing and the many rare disease therapies is understandable, it must be compared to the costs on the health system for patients who are not diagnosed and who do not receive proper treatment.

Dr. MacKenzie will speak more about the costs of treatment, but these costs do not grow linearly with the number of patients. Because so much of the cost of a drug is sunk research costs, the more patients identified brings down the per-patient costs, while the benefits multiply proportionately.

The key to this is identifying and aggregating information for all rare disease patients. Canada is a small country in terms of population, and people with a rare disease are spread all across it. Our efforts on precision medicine in rare diseases, including early genetic testing, have to be national in scope.

In summary, I would like to make three recommendations to the committee.

First, when considering the costs of treatments and interventions, we always think about the aggregate costs and also the aggregate benefits at the economic, societal and personal levels.

Next, genetic testing for people with rare and/or undiagnosed diseases needs to be given a priority in any strategy that is aimed at removing barriers to access to treatment. This testing has been proven to reduce overall costs and can open the door to therapies, as it did for Ian.

Finally, because so few people have any individual rare disease and higher numbers significantly reduce costs, a national strategy to address access to rare disease drugs in particular and precision medicine in rare diseases more broadly needs to be national in scope.

Thank you. We'll be happy to take your questions.

● (0835)

The Chair: That was six minutes and 54 seconds.

Thank you for that personal testimony. That means a lot to us. It helps us to get a grasp on this.

Now we'll go to Dr. Alex MacKenzie.

Dr. Alex MacKenzie (Clinician Scientist, Children's Hospital of Eastern Ontario (CHEO)): That's a wonderful job, guys. That's a hard act to follow.

I'm a clinician scientist at CHEO. I work on rare diseases. In deference to the Dutch, I'm going to keep it pretty brief and open it up for more questions.

I would like to run the numbers. This is probably old news to the committee but I'll give you a numerical overview.

The number of rare disorders is 7,000, as Michael said. That number may well grow with time but that's our estimate right now. Roughly one million Canadians are affected. One in 12 is bandied about. We think that is an overestimate. It's closer to 2% to 3%. Nonetheless roughly one million Canadians are affected.

Over 50% are children. Under 50%—less than a half— have a diagnosis now. When I look at the in-patients on my ward at CHEO, I see that roughly one-third have rare diseases. These diseases are responsible for one-third of deaths in the first year of life, and one-third of children with rare diseases will die before they reach the age of five. That's to give you a sense of the impact of rare diseases.

The proportion of rare diseases for which there is no therapy is 94%. We do not have a therapy for most of these disorders.

Perhaps the most telling statistic is that when you look at the proportion of general years of life lost for rare diseases, it's around 4.6%. That's years of life lost in Canadian society. For infectious diseases, the number is around 1.4% or 1.6%. For diabetes, it's only 2.6%. It's really dramatic. I think the reason for this is that it takes life early on, so that carries a disproportionate impact.

As Michael said, we live in revolutionary times right now with economic DNA sequencing. The first responders to this are genomes. Everybody in this room will be in our electronic medical health record within a decade—no question. It's going to impact how we're perceived at risk of disease, prevention and therapeutic options. Whether you call it genomic, precision or personalized medicine, it's a revolution.

The children with rare diseases are really the first responders to this revolution. I would say parenthetically that getting this right is not just good for children with rare diseases; it's good for all of us.

We have in the guise of Michael, whose Matchmaker Exchange platform has been adopted by Johns Hopkins in Baltimore, Cambridge University and the National Institutes of Health in Washington, an absolute global leader in rare disease diagnosis. He's too modest to say so.

With Kym Boycott, my colleague leading Care4Rare at the children's hospital, we have a pan-Canadian consortium where we've diagnosed rare diseases at an unprecedented rate—almost 200 new rare disease genes identified. There's no other developed country that has really done a national, full-on assault like that. I'm not sure if it's the submersible Canadian ego, our desire to work in committees, but we really are number one as far as going coast to coast to coast and addressing this problem.

As we heard so eloquently from Ian, a diagnosis really is a form of therapy—absolutely critical. When Mark LePage talked to you about their Genome Canada initiative to do 30,000 rare disease genomes, that's a tremendous project with huge potential. It's a bit like Dickens: the best of times and the worst of times. It's incredibly exciting what's going on as far as diagnosis is concerned, but the lack of therapies and also the cost of therapies must be mentioned.

My laboratory works on repurposing drugs. We take clinically approved drugs and look at indications for rare diseases. In working over the last five years, we've identified five potential therapies for diseases involving epilepsy, aortic aneurysms, nerve degeneration and muscular dystrophy. These drugs cost as low as a dollar a day, sometimes less than that.

Dr. Vicky Siu, a colleague in London, is giving histidine to the old Amish individuals with a seizure disorder. Clara van Karnebeek, at UBC, has started a web page to show doctors how to use dietary modulation for rare diseases. There are other ways of addressing rare diseases than new drugs.

But obviously new drugs will be needed and therein lies the rub. What can Canada collectively do about the problem of the cost of these drugs?

● (0840)

Mike laid out very well the economic argument for looking at them perhaps differently from other drugs.

I would just say a few things. I think we need to show a united front. Right now, Canada is alone in the OECD and the developed world in having a balkanized provincial approach. This is shown in how, after Switzerland and the U.S., we pay the third most in the world for these drugs. We have the pCPA, but we really need a stronger policy lever in this regard. We need to start prescribing biosimilars and generics more than we do now. These are under-prescribed.

The U.S. introduced legislation about 15 years ago to accelerate biosimilars. These are drugs that are the biologicals, the antibodies, etc., that aren't exactly the FDA-approved drugs, but are the generic form. This legislation in the States has made a difference.

We need to look at things such as managed access programs, where the companies generate data as we go along with the therapy so we can assess who should get their drugs, based on hard evidence, and who should actually stop the medication. It is one of the most difficult things to face—at what time do you actually stop a medication when it's not being effective?

For my part, I think we may need to rethink how we go after rare diseases. Right now, it's academics like me working in labs collaborating with big pharma and biotech companies, but I think we need the almost Thomas Edison or Henry Ford plant approach to rare diseases in a generalized fashion, where you have open access, much like what Aled Edwards talked about. We need a transparent factory approach where you're making gene vectors, which go to specific tissues that you can put different genes into, novel ways of isolating proteins in an industrial fashion, a really systematic approach to repurposing drugs such as what we did, the idea of a universal donor cell where you can actually generate cells that do not induce immune responses, such as those that Jonathan Pitre, the butterfly boy, succumbed to just last year.

There are a number of thoughtful systematic approaches one can use to tackle rare disease therapy generation.

The Chair: I have to ask you to wind up, there.

Dr. Alex MacKenzie: Yes, and that's it. I just want to say that—

The Chair: You wound up; that's perfect.

Dr. Alex MacKenzie: —basically, it will sort itself out one way or the other, but I think we need to tackle this problem.

The Chair: Thank you very much.

Now we go by video conference to Dr. Lexchin.

Also, I should point out that some of this will no doubt be in French, so we have interpretation services available. You will need them eventually.

Okay.

Dr. Joel Lexchin (Professor Emeritus, School of Health Policy and Management, York University, As an Individual): Thank you very much for the opportunity to appear. I'm giving this presentation not just on my behalf but on behalf of the other 10 people who signed the written brief.

This presentation is going to concentrate on drugs for rare diseases, and I'm just going to go through the recommendations that we have.

First of all, if there is a policy around rare diseases, it should not be based on the assumption that drugs are not commercially viable just because there are small numbers of people who have the disease. In order to prove that our research and development into drugs for rare diseases is not commercially viable, companies should have to divulge their R and D costs and show that these costs are not commercially acceptable, given the number of people in Canada that the company expects to treat with the medicine and the price it plans to charge.

Second, based on U.S. figures for the number of drugs for rare diseases that have been approved recently—and recently in the past half-dozen years or so, 37% of drugs in the U.S. have been labelled as drugs for rare disorders—there may not actually be any need for Canada to provide monetary incentives for research into rare diseases. However, it may be appropriate for Health Canada to set up a distinct regulatory path for these drugs. As an alternative to providing companies with monetary incentives, the Canadian government should consider investing more money into research into these diseases.

Third, if the overall prescribing rate for an orphan drug exceeds the number of people with the rare disease, then the drug should lose its orphan status.

Fourth, when more than one disease is caused by the same pathophysiological mechanism, and therefore could be treated but with the same drug, then each disease should not be considered separately when deciding whether a drug should receive orphan drug status.

Fifth, Health Canada needs to guard against the acceptance of biomarker subsets of disease and limit the use of the orphan drug designation to situations where the drugs are truly distinct. For example, if the same drug treats more than one genetic abnormality that causes the same disease—so we have a drug for cystic fibrosis that's being used to treat cystic fibrosis caused by two different genetic mutations—then each cause of the disease should not be treated as a separate rare disease.

Sixth, while small numbers of people with rare diseases place limitations on the design of clinical trials, Health Canada should demand the highest degree of rigour possible in these trials. In addition, post-market clinical trials should be a requirement when the evidence regarding either clinical benefit or safety is unclear, and Health Canada should monitor and publicly report on the progress of these post-market trials.

Next—and this is a point that's already been made—the assumption that one in 12 people have a rare disease in Canada is unreliable and should not be used to form Canadian policy about drugs for rare diseases. Any definition of a rare disease should not just take into account how frequently it occurs, but also needs to incorporate the element of severity. In other words, it should occur infrequently and also be severely debilitating.

Finally, any recommendation that the House of Commons standing committee makes based on the testimony of groups representing patients with a rare disease should take into account conflicts of interest that these groups may have with companies that would benefit from any policy that's developed.

Thank you.

● (0845)

The Chair: Thank you very much. I appreciate your concise comments.

Now we'll go to Janssen Inc., Pharmaceutical Companies of Johnson & Johnson, for seven minutes.

Ms. Stacey Silverberg (Stakeholder Engagement Manager, Government Affairs and Market Access, Janssen Inc. Pharmaceutical Companies of Johnson & Johnson): Thank you.

Honourable members, I would like to thank the committee for inviting me here today to speak with you regarding your study into barriers to access to treatment and drugs for Canadians affected by rare disorders. As I begin, I would like to share a bit of background as to why I've been afforded this particular opportunity to address all of you today.

I work for Janssen, which is part of the Johnson & Johnson family. I have worked with them for four years; however, I do come to you today with 25 years' experience in the innovative pharmaceutical industry. While I've had a variety of roles through my long and rewarding career in this sector, I'm so incredibly grateful for the time spent meeting patients, working with patients and being part of their journeys, especially when I've been able to personally witness the impact we have had, and will continue to have, on their overall survival, their quality of life and, yes, even in some cases, their cure.

Over my tenure in this industry, I have been able to observe patients with schizophrenia reintegrate into society and live more productive lives than they or their carers ever could have imagined. I've stood witness to cancer patients being put on a new treatment that has literally breathed new life into them. In fact, cancer can now be considered in some cases a chronic disease versus a death sentence, once again because of access to innovation and because of this innovation. These are just a couple of examples of some types of patients I've had the privilege to work with over the last many years.

Today, I would like to ask all of you to ensure that we don't do anything to unintentionally put barriers in the way of these patients, and truly look for ways to additionally facilitate their ability to access medications as one part of their treatment plan. I would also ask that you look to afford the same type of access to innovation for patients living with rare diseases, a very vulnerable population.

I will share with you a brief overview of Janssen's work in providing access to innovative medicines, specifically focusing on the rare disease population. I will speak to the need for an orphan drug regulatory framework, which should be designed to enable development and availability of medicines for rare disorders. Finally, I will provide an overview of concerns we have with two ongoing government initiatives that we believe may limit access to new innovative drugs for rare diseases.

In Canada, as you may be aware, we don't have a formal rare disease framework or strategy, but it is important to note the “one in 12” number that's been noted today. As well, that number comes from the Canadian Organization for Rare Disorders. One in 12 Canadians lives with a rare disorder. It is incumbent on us, as Canadians, to help this at-risk population.

Back in June, 2017, Johnson & Johnson acquired Actelion, a leading innovator in the rare disease pulmonary arterial hypertension, or PAH population. PAH is a rare disease that I will address today. For further details, in the interest of time, please refer to the written documentation we submitted to you earlier.

PAH is a progressive disease that can strike at any time and does not discriminate. It is a challenging disease to diagnose, and often these patients languish in the system. The smallest everyday tasks that you and I take for granted seem to a PAH patient to be like climbing Mount Everest. As mentioned, it's a progressive disease and it can lead to fatal consequences. If left untreated, these patients live an average of two to three years from the time of diagnosis.

Although there is no cure for PAH, there are treatment options, and due to the innovative medicines approved in Canada, Canadians can live longer and healthier lives. These treatments have meant that now 50% of patients survive five years or more from time of diagnosis, but access to these medicines is limited, often with criteria to meet and delays in approval. Some are not even reimbursed by public payers. These are crucial medicines for those who need them and delays to access can be devastating.

A strategy for rare diseases would help address these issues, better defining what treatments should be made available to patients, to ensure there is funding available to pay for them.

There are barriers to access that arise because of uncoordinated and uneven approaches across the country to both the regulation and reimbursement of these medicines. To address this, Janssen encourages the federal government to implement an orphan drug regulatory framework that would offer additional support to encourage the development and availability of orphan drugs in Canada. Recent government efforts, including the orphan drug regulatory approach, are very encouraging, but this framework would further solidify the government's approach to accessing drugs for rare diseases.

Additionally, given there are uneven and unequal funding approaches to drugs for rare diseases across the country, Janssen encourages the federal government to fund a separate drug program specifically for rare diseases. This could be done in parallel with the ongoing work related to national pharmacare, and would ensure that funding would be available when Canadians need these medically necessary medicines.

● (0850)

In the same vein, we are concerned that two ongoing initiatives of the federal government could further exacerbate these issues of access: the proposed reforms to the Patented Medicines Prices Review Board, PMPRB, and the implementation of national pharmacare. We recommend for both initiatives that the federal government closely consider the potential implications to ensure that these do not have unintended negative consequences.

Regarding the PMPRB changes, we are concerned that the new regulations, as they stand, could decrease access to new innovative drugs, including drugs for rare diseases. The updated regulations may result in Canada not being an early-launch country for drugs, which would slow access to innovative medicines that Canadians need. It has been seen that fewer medicines are launched in countries with prices at the median of the new proposed comparator countries, including medicines that target rare diseases.

We encourage the federal government, as it develops a national pharmacare policy, to consider potential implications for Canadians with rare diseases. Coverage for new innovative medicines is essential to ensure the best care for rare diseases, but moving to a single-payer public plan may impede this. Accessing new medicines is time-sensitive for patients with rare diseases. It is vitally important that the existing public and private mix of drug plans be maintained to ensure that the latest medicines are available for Canadians.

Finally, both changes could result in fewer clinical trials in Canada. For those with rare diseases, and especially life-threatening illnesses, clinical trials are the earliest means to getting innovation and hope to patients. In Canada we value health. In Canada we value safety. Let us ensure that these proposed policy changes do not put at risk some of the core values that define this wonderful country.

This study you have so boldly undertaken can have a substantial impact on three million Canadians. As you continue these deliberations, I urge committee members to take a close look at concerns raised by me and others regarding the PMPRB reforms and national pharmacare to ensure that the implementation of these programs does not have unintended negative consequences that could limit access to new innovative medicines for rare diseases.

I would like to thank the committee for providing Janssen with the opportunity to speak today. It has truly been an honour.

● (0855)

The Chair: Thank you very much.

Thank you, all, for your opening remarks.

Now we'll go to our seven-minute questioning round, starting with Mr. Grewal.

Mr. Raj Grewal (Brampton East, Lib.): Thank you, Mr. Chair.

Thank you to the witnesses for coming here today. I'll be splitting my time with the honourable member to my left.

Dr. MacKenzie, you put the number at around one million, and everybody else, including the documents I have, puts it at about 2.8 million Canadians suffering from rare diseases.

Can you elaborate on why your estimate would be 1.8 million?

Dr. Alex MacKenzie: That's roughly 2% to 3% up. It's just based upon our personal observation at the CHEO genetics clinic and extrapolations therefrom.

If one drills down in the literature—and perhaps Joel might have a comment, because both he and I are saying that one in 12 is perhaps a bit inflated. That in no way is to undermine the severity or seriousness of rare diseases. I just think we need to be as careful as we can about the numbers.

If you look at Australian, Belgian and Italian studies, in which they have done this well, it comes in at roughly that benchmark. Given the diaspora that makes us up, there is no reason to anticipate, from a genetic point of view, that it would be any different from the 2% to 3%.

I think there may be a bit more data. I tried to drill down on the one in 12 from the CORD web page, but I am unable to find the source of those numbers.

I'm not sure if Joel—

Mr. Raj Grewal: I think if you want something that everybody on the panel can agree with, identifying the number of Canadians suffering from rare diseases is probably the number one priority in order to address the problem.

Dr. Alex MacKenzie: I would agree, and I think if we go ahead with the Genome Canada project to do the 30,000 rare disease genomes, that will be a step in the right direction, as would be other general genomic sequencing projects.

Mr. Raj Grewal: Thank you.

Ms. Silverberg, thank you for your testimony. You work for Johnson & Johnson, which is a large pharmaceutical.

You mentioned that in Canada we value health and we want to ensure a healthy population. I think the job of any government is to ensure the success of its people.

Isn't it a bit of a conundrum for Johnson & Johnson that you're developing pharmaceuticals and your number one objective is for shareholder value? That comes in contradiction with developing drugs that would be helping people with rare diseases, because some of those drugs may not be profitable in the market but would do good in terms of the social licence associated with them.

Could you comment on Johnson & Johnson's role as an important stakeholder in developing a solution to this problem?

Ms. Stacey Silverberg: Johnson & Johnson is actually bound by our credo. It really helps us with every single decision that we make. Our credo puts patients, as well as doctors and nurses and mothers and fathers, at the core of everything we do. That's our guidepost for everything that we look to do. Ultimately we are responsible to shareholders as well, but there's a huge opportunity to have a mix, and I do believe very strongly that we do that. We are a for-profit organization, undoubtedly, but we are patient care first and foremost.

I have been privy to some recent meetings where we know things will not be as commercially viable, in some of the terminology used today, but again, going back to the credo, it's the right thing for us to do. If we have new innovation to bring to patients, we need to continue to work in that vein and put patients at the centre of every decision we make as an organization.

● (0900)

Mr. Raj Grewal: Thank you.

Doug.

Mr. Doug Eyolfson (Charleswood—St. James—Assiniboia—Headingley, Lib.): Thank you very much, and thank you all for your presentations.

Ms. Silverberg, something that Dr. MacKenzie said echoes what we found repeatedly during our pharmacare study, that Canada pays the third highest cost for medications in the world. You're concerned that changes to the PMPRB could decrease access to innovative drugs. How is it that the rest of the world is paying so much less for drugs, but putting our country more in line with the rest of the world would cause this decrease in innovation?

Ms. Stacey Silverberg: It is a difficult issue to understand, and admittedly it took all of us in the community who are not health economists a while. I don't profess to be a health economist. My background is certainly coming up through the pharmaceutical organization.

What we understood going forward was that, when you look at the current basket of countries, Canada has a favourable position whereby we get early access because of the pricing and the comparative countries being used. With the new proposed changes, that does put us in a situation where a lot of lower-cost countries are put now in the comparative basket, compared to where we currently are with the seven countries being referenced. Because of that, what it actually means for patients, and all future patients in Canada, is that, with a less favourable pricing regime, this will potentially affect the opportunity to stay an early-launch drug country.

Secondly, it may not foster an environment where global companies will look to Canada for clinical trials. That is the reality of being 2% of a global picture, that Canada unfortunately may be at risk for not bringing that innovation, starting at its earliest point, here, which is the earliest point for people such as Ian and his daughter to have access to new treatments that may help them.

Thirdly—

Mr. Doug Eyolfson: Sorry to cut you off, but I have limited time.

For the common diseases such as diabetes, insulin is a century-old drug and the price hasn't gone down in literally decades. It's still quite inflated. The prices of life-saving medications such as EpiPens are actually skyrocketing, despite the fact that it's a fairly basic drug.

Are you saying, in order to foster the development of medications for rare diseases, we have to keep common drug prices artificially high?

Ms. Stacey Silverberg: I don't think they're artificially high.

There are two things. One, we are heavily regulated to ensure non-excessive pricing. There is the PMPRB, of course, and that is the list price. We also have the opportunity to negotiate. As many of you are aware, we have confidential listing agreements.

Going back to access, affordability—

Mr. Doug Eyolfson: Sorry, I need to cut you off. I have the answer I want.

I have one last question. How will a single-payer plan decrease access as opposed to a public-private insurance plan for which you're advocating?

Ms. Stacey Silverberg: Public plans don't list drugs as quickly, nor do they have the breadth of what private plans list for employees who are afforded that opportunity to get them through their employer. The mix is actually making sure that we have availability today.

Thus, a future pharmacare program should ensure that we don't have less access than we do today. The mix will ensure that we cover those hopefully going forward, both publicly and privately, and that is a proposed solution that we feel would be best for all Canadians, to make sure everybody has an opportunity to be insured and at the highest possible level they can be.

Mr. Doug Eyolfson: That's my time. Thank you.

Ms. Stacey Silverberg: Thank you so much.

The Chair: Okay, now we go to Mr. Kmiec.

Mr. Tom Kmiec (Calgary Shepard, CPC): Thank you, Mr. Chair.

Let's start with Janssen. I have a rare disease in my family called Alport syndrome. It's a very personal thing for me. I guess my family would not be the ones you would think of. I thought genetic diseases were about people who were interrelated having kids. My wife is from Singapore and I'm Polish, so there's absolutely no way our ancestors are connected in any way.

On rare diseases, drug access is the biggest issue that I see. Too often I see people talk about the sticker shock—that was mentioned in the presentation—of seeing the final price for a particular rare disorder. The comparison I make is that I see sticker shock whenever I see a Lexus or BMW, but what I don't see is the bargaining agreement, the plant, the investments in dollars, the researchers, in the case of drugs. All of that cost is baked into it. Part of that is also clinical trials.

You're a company that does clinical trials. How long is it taking, and what are the costs baked into it? That portion of it has a big impact on the final price tag, and then there are the negotiations that you do with the different provinces and pCPA and the private companies as well.

Can you talk about that cost structure?

● (0905)

Ms. Stacey Silverberg: I wish I could. Unfortunately, I don't have the in-depth knowledge about what goes into our clinical trials.

First of all, let me just say I hope that your children are doing well living with the rare disease. I hope that innovations have been able to give them a quality of life they deserve. A lot of that is due to the work done by the innovative pharmaceutical industry, so people like you and Ian and his daughter actually have the opportunity to live fruitful lives here in Canada.

In terms of research and development costs, they are significant and that doesn't account also for so many failed molecules that never actually make it to market.

Mr. Tom Kmiec: What's the ratio on that?

Ms. Stacey Silverberg: I actually don't have that information, but I'd be happy to follow up.

Mr. Tom Kmiec: Could you submit it to the committee?

Ms. Stacey Silverberg: Absolutely.

Mr. Tom Kmiec: Thank you, and if you have any information about the economics that go into it, that would be great too.

Mr. Brudno, you're the one who mentioned sticker shock. Drug access is the big portion of it. It's really led by a lot of patient groups. When my kids were diagnosed with Alport syndrome, it was after a lot of misdiagnoses. When you talk about early diagnostics, I totally understand.

I met my physician by accident at a patient medical conference in Minneapolis. He was standing right behind me when I asked if there was a Canadian there. He happened to be the head of pediatric nephrology at the Alberta Children's Hospital. I think he was one of the gentlemen who presented at the committee. I met him wholly by accident. Had I not done that, I would have gone through a lot of misdiagnoses.

I'm sure, Mr. Stedman, you went through the exact same thing I did.

We got to the point where the federal government had actually created a rare disease framework to make it easier to provide a pathway for rare disease drugs. That was killed off. The court called it the kiss of death and they got rid of it. The federal government didn't continue with it after 2015. It kind of laboured, and it's gone now.

What would a rare disease framework look like to you specifically in the government? The FDA has kind of a fast-track process for the approval of drugs, but there's approval of drugs, there's drug access and then there's reimbursement as well. I have met a lot people in my riding who have rare diseases I have never heard of. Their drug is approved in Canada but not for reimbursement. They face huge out-of-pocket costs because their public insurer refuses to cover the drug.

Can I hear from you on that?

Dr. Michael Brudno: In general, I think it's important when we look at.... All of these are important components: having the access, having the drug approved in Canada through the efforts of pharmaceutical companies doing clinical trials. Then making it available means making it reimbursable to the extent possible within the public health care system.

Out-of-pocket costs for these drugs can be huge and they are not bearable by individuals, but they can be borne by a society that averages those costs across lots of other individuals and also a society where we identify as many patients as possible who are eligible for a specific drug, which will cause the prices of those drugs to go down.

I don't think I can give you a specific sort of recipe—this is what you do here, this is what you do there. Obviously everything in the health economics space is a question of balance. You're balancing the costs to the health care system versus the benefits that this brings to all of the individuals who are affected and to the society at large.

Mr. Tom Kmiec: Can I interrupt you?

I just wanted to mention, on the cost of pharmacoeconomics, the role of patients in this. Don Bell was a great New Democratic MP. I'm going to say that; hopefully Mr. Davies is listening.

Dr. Alex MacKenzie: It's on the record.

Mr. Tom Kmiec: Don Bell had a grandson who was three when he was diagnosed with pulmonary hypertension. It was a terminal illness for him. Don made it his case. There was a debate in the House. There was a motion put forward, but for him, it was a patient issue. Too often I find we develop national research policies, all these grandiose schemes, but at the end of the day, it's the patient who really cares and is really motivated.

People like Erin Little, people like Roy Vinke in my riding and people who really care about SMA are the ones who create these little foundations, and they start pushing pharmaceutical companies to do research. They find a diagnostics company. The diagnostics company for Alport syndrome happens to be in the United States. There's no lab in Canada that can do the test.

The role of the patient in this and patient-centred care, I feel, are lost when we start talking about big single-payer national pharmacare because it's going to be about rationing. Reducing costs can only happen through rationing when you have an expensive drug.

Can I hear your comment on that?

Dr. Michael Brudno: The role of the patient is huge. I know of an example of a someone in the U.S. who went from having a child who was the first in the world diagnosed with a specific rare disease—literally the first—to creating a patient group of now around 50 patients worldwide, all within the span of five years, to actually becoming a professor of medicine within the span of the same five years. He was a professor of computer science—so my colleague—and he actually started to create therapies for his son's disease, all in five years, I believe.

Patients have great power to enact change. Yes, there is a balance between the two. Patients can't do everything. They need to work with pharma. They will need to work.... The key to me is to identify early as many of these individuals as we can, making sure that they don't fall through the cracks and that the diagnostic odyssey stops as early as possible so that patients can start working on the treatment side.

● (0910)

The Chair: Mr. Davies.

Mr. Don Davies (Vancouver Kingsway, NDP): Thank you, Mr. Chair.

Thank you to all the witnesses.

Dr. Lexchin, I'd like to start with you, please. In an article that you authored in Maclean's in March 2018, you wrote the following:

> While there are drugs that are not sold in Canada, the reason is the relatively small Canadian market, not the price.
>
> In fact, when it comes to paying for prescription drugs, only the United States and Switzerland outspend Canada on a per capita basis out of 31 industrialized countries in the Organization for Economic Cooperation and Development.

Do jurisdictions that currently pay lower drug prices than Canada, such as France or the U.K., face slower or more limited access to new life-saving medicines and vaccines than Canada?

Dr. Joel Lexchin: No, they don't. First of all, let's be clear. Based on objective studies, only about one in 10 new drugs that are introduced in any given year make a substantial difference to the therapy that people get. Over half of the drugs that are introduced are actually what are sometimes called me-too drugs. In other words, they're an attempt to get into the market, but they don't offer any additional value.

As far as how quickly countries access the drugs, that largely depends on the size of the market. Canada, as the representative from Janssen said, is 2% of the worldwide market. The United States is about 40%. Some European countries are 10%. Companies are going to go to those markets first because they start to get money back earlier on. Canada is later on. In fact, companies wait about six months longer to file for approval in Canada compared to the United States.

Mr. Don Davies: I'd like to follow up on the elephant in the room when you talk about rare diseases and prescription response: price. You also wrote in that article, “There are now 19 drugs on the Canadian market that cost $50,000 or more per year, compared to just six a decade ago.”

You used the example of a life-saving drug to treat cystinosis, a rare disease affecting probably 100 people across Canada. When you said that it's "soon to rise from $10,000 per year to more than $300,000 annually", you indicated that the new form of the drug "contains the same active ingredient as the old form of the drug", but that "it differs only in that it contains a new coating, enabling a slower release of chemicals into the body." You pointed out that "the basic research and development...was financed by patient groups, not drug companies", and that "Horizon Pharma has not publicly offered any reason for the price it plans to charge."

What can you tell this committee about our need and our desire as policy-makers to make sure that Canadians suffering from rare diseases get access to significant new developments, and how do we measure these massive costs with efficacy?

Dr. Joel Lexchin: You've hit upon an issue that people have been talking about for a long time: that the drug companies will not open up their books to reveal their R and D costs for new medications. There's a figure of $2.6 billion that's bandied around as being the cost of getting a new drug to market. That kind of figure is based on confidential data that won't be released. If drug companies want to prove that they need to charge these significant amounts of money that they do for new drugs, then they should prove to Canadians and to insurers that those prices are actually justified. However, so far, they haven't.

When you look at some of the other countries.... For instance, you were pointing out that Canada is just behind the U.S. and Switzerland. In Denmark, where there aren't any bodies lying around on the streets due to lack of drugs, they spend $240 per person per year on medications versus over $700 per person per year in Canada.

● (0915)

Mr. Don Davies: What about the efficacy issue? How do we decide whether or not it's worthwhile to publicly fund a drug if the impact on it is maybe only marginal? How do we make that tough decision?

Dr. Joel Lexchin: Those decisions are never easy. Often, actually, when drugs for rare disorders come on the market, because of the small numbers of people, we really don't have enough information about those products to make good decisions. That's why we need ongoing studies once those drugs are on the market.

As Dr. MacKenzie, I think, was pointing out, if those drugs, based on the ongoing studies, prove not to be beneficial either to people as a whole or to individuals, we need to be prepared to stop paying for those. Obviously, if they're beneficial, then we should continue to pay for them.

Mr. Don Davies: Dr. MacKenzie, you mentioned, and I'm sorry if I got this wrong, a 30,000 rare disease genomes project.

Dr. Alex MacKenzie: Yes.

Mr. Don Davies: How can the federal government support it?

Dr. Alex MacKenzie: How can they support it? Marc LePage is here as an observer, and perhaps he could elaborate later on.

Fundamentally—not to go into some inside baseball talk here—it's in the budget this go-round, I think, for part of the global Genome Canada ask. That's how it's going to be remunerated, and I just think it's overdue. As Michael said, there are five million U.K. genomes being done right now. We're asking for 30,000. We're international leaders in this realm, and I just think the return on investment will be phenomenal.

Sorry; that's a little subjective.

Mr. Don Davies: Thank you.

Ms. Silverberg, I'll give the last question to you. If I heard your testimony correctly, you suggested that there's a relationship between early launch and the impact of pricing. I think you suggested that Canadians may risk having early launch of drugs if we reduce prices. I wrote down your words, and you said that a "less favourable pricing" environment may result in reduced access.

If, as a drug company, you're putting patients first, why would that be?

Ms. Stacey Silverberg: The reality is that we do put patients first. We will do everything to try to make things available, and they will become available. The timing of such a decision may potentially be affected, though, because we are a global organization, and there will be countries that will have an opportunity, again, as patients, to have it earlier.

We are worried about our Canadians. They will eventually potentially get access, hopefully in most cases, but there will be a risk of not launching here, because ultimately decisions have to be made as to what's best for other patients in other jurisdictions. We never want to put patients at risk.

The reality is that those who are lower than the OECD median.... In fact, PMPRB creates their own reports, so the data is actually there. New countries, such as the Netherlands, are being entered into the comparator basket. New Zealand is not in the comparator basket, but only 16% of drugs are being launched there because of an unfavourable pricing structure. In Spain 21% of drugs are being launched.

Right now, with the current environment, 50% of drugs in Canada are actually being launched here because of the prices that are afforded within this—

Mr. Don Davies: That sounds like putting pricing first.

The Chair: Sorry, your time is up.

Now we go to Ms. Sidhu.

Ms. Sonia Sidhu (Brampton South, Lib.): Thank you, Chair.

Thanks to all of you for being here.

My questions are for Dr. MacKenzie and Dr. Brudno.

You said that for neonatal testing, 50% of kids died before the age of five. How can we get to a solution for genetic testing in terms of neonatal testing?

Dr. Alex MacKenzie: Michael, I'll tackle it first.

Basically, I think clinically introduced DNA sequencing will identify these individuals before they manifest their symptoms. That's the short answer. We have newborn screening in our institution where we look at 150,000 babies a year. For Ontario we study 40 diseases. With new exome sequencing, in the brave new world we'll be able do for it hundreds and thousands of disorders, to identify those pre-symptomatically.

● (0920)

Dr. Michael Brudno: To follow on from that, I would slightly disagree with my colleague in that I think pre-symptomatic testing using genomics is still a little bit further away. However, the ability to have very rapid testing once the symptoms onset can help improve outcomes significantly.

In the U.S., a 48-hour genome is becoming a new norm in neonatal intensive care units. When you have a baby who's extremely sick, they will bring a genome. They will do a test. They will try to identify the right therapy within 48 hours. That's instead of trying option one, option two, or option three, during which time the baby is getting progressively worse and during which time there is a deterioration that may not be reparable once the right treatment is identified. With certain seizure disorders, a baby's brain will basically fry before the medicine has the ability to kick in. You need to find the right medicine and you need to find it quickly.

These very rapid testing regimes, which the precision medicine initiatives like the ones suggested are trying to bring to fruit, are the right approach.

Ms. Sonia Sidhu: Thank you.

There's a lot of potential for artificial intelligence to help people living with rare diseases and disorders.

What are the major advantages of artificial intelligence? Can you explain that? Ian, can you explain your thoughts on that too?

Dr. Michael Brudno: Artificial intelligence is another way of saying “really advanced computing”.

There are advanced computational tools that can help individuals in all phases of our lives. With your Siri assistant or your Google Assistant, you can do things by talking that you previously would have had to spend a lot longer time typing or entering on the phone.

It's the same in the case of rare diseases. These artificial intelligence methods can help patients undergoing therapies who need help with day-to-day tasks, which they can get artificial intelligence to do for them.

At the same time, artificial intelligence also forms the core of how we analyze these genomes. Having large numbers of genomes allows us to learn from these, to identify what changes in the genomes correspond to what clinical outcomes, and to identify why with two individuals with exactly the same genetic mutation, one may be playing soccer and the other is on a ventilator and unable to walk.

Having more and more patients, more and more data, are critical for artificial intelligence solutions, to learn the difference between those two patients and to be able to predict, for new patients, the most likely trajectory and the best intervention.

Dr. Alex MacKenzie: We also use it for therapeutic configurations for rare diseases as well. We identify potential therapies without actually going to the benchtop and test tubes. We're just doing a sort of computational analysis.

Ms. Sonia Sidhu: Thank you.

Ms. Silverberg, with regard to PH or pulmonary hypertension, is it considered a rare disease?

Ms. Stacey Silverberg: Yes, it actually is defined as a rare disease.

Ms. Jacqueline Dobson (Government Affairs and Policy Manager, Government Affairs and Market Access, Janssen Inc. Pharmaceutical Companies of Johnson & Johnson): For the record, it's pulmonary arterial hypertension, which is otherwise known as PAH, for short.

Ms. Stacey Silverberg: This is the rarest form of pulmonary—

Ms. Sonia Sidhu: Do you think that when people are enrolled with clinical trials that big trials are more effective?

Sometimes all the population cannot participate in clinical trials. How do you measure the efficacy for the trials?

Ms. Stacey Silverberg: That would not be my area of expertise, but we'd be happy to follow up for you.

Ms. Sonia Sidhu: Anyone else?

Dr. Alex MacKenzie: Big trials aren't possible. You need to go to even one trial sometimes because it is so rare. You just need to be very fastidious about the biomarkers and that the questions you're asking are doable.

It's a new way of doing clinical trials, no question.

Ms. Jacqueline Dobson: Clinical trials are not just one phase. They occur in three phases.

As I'm sure you can imagine, progressing to each phase becomes more and more difficult, even before it can get going through our pricing regulations, as well as through our commercialization bodies through the provinces to be listed.

Ms. Sonia Sidhu: Thank you.

The committee heard from Dr. Craig Campbell, from his department of pediatrics and neurology at the Children's Hospital at the London Health Sciences Centre, that registries are essential to capturing data on the health benefits and risks of new drugs on rare diseases.

Can you describe what additional steps Health Canada could take to support the surveillance of the safety and efficacy of a drug that could be more effective?

Dr. MacKenzie.

● (0925)

Dr. Alex MacKenzie: Craig Campbell, an esteemed colleague, has a Canadian neuromuscular disease registry that is run out of Calgary. It is a very impressive disease registry, where you can track safety and do clinical trials. It's a perfect model for how things should be done in a perfect world.

I think as we go ahead with the companies and new drugs being introduced, an important part is setting up these networks and capturing the data as we go forward, ensuring efficacy and ensuring safety. I know the that the CNDR is in conversation with Biogen on spinal muscular atrophy, for example.

I think that's an absolutely important aspect that we need to do, ultimately not to sort of perseverate with the Genome Canada project, but the introduction of these rare disease genomes is going to be a good platform from which to move on to registries as well.

Ms. Sonia Sidhu: Is there any central data for that registry?

Dr. Alex MacKenzie: It's a big question, and Mike spends a lot of time looking at this.

With regard to the genetic sequence for sure...and on the actual description of the disease, there's something called the HPO, the human phenotype language that Mike uses.

Do you have anything to add?

Dr. Michael Brudno: I would just mention that a lot of these registries are driven by either researchers or clinicians who have information.

There needs to be much more of a push to get more information directly from patients. Information coming straight from patients is obviously going to be in some cases less reliable, but there are going to be aspects of a disease that a clinician never sees, things that happen only at night, or things that affect the patient significantly but don't make it into what they discuss with their clinicians.

Mr. Ian Stedman: Yes. For example, I have seven different symptoms associated with my disease, but I never put them together. Growing up, I would talk about one or the other when I went to see my doctor, or about whatever was bothering me that day or that week.

If I could record what was happening to me on an ongoing basis and that would be somewhere, like in a registry, with other people just like me, who knows...?

Ms. Sonia Sidhu: It can be helpful for the other patients too.

Thank you.

The Chair: The time is up, and I'm sorry it is because I feel that we've just scratched the surface of the knowledge that you can impart to us on this issue.

Dr. Alex MacKenzie: We are very eager to.

The Chair: Dr. MacKenzie, you wanted to make a comment on Mr. Kmiec's question and I missed it. You signalled that you wanted to make a comment. I don't remember the question, but if you remember the question, you're certainly welcome to comment.

Dr. Alex MacKenzie: I think I was just twitching, perhaps. I don't know.

Voices: Oh, oh!

Dr. Alex MacKenzie: I do want to say that the calibre of questions was very impressive. There was real thought brought to bear.

The Chair: The quality of our committee makes my job very easy, it really does, as does the quality of our witnesses. Our members do their homework. It's the most productive committee, I think, and we have the best witnesses.

Ms. Marilyn Gladu (Sarnia—Lambton, CPC): Hear, hear!

The Chair: I want to ask Mr. Stedman a couple of questions.

Your daughter was diagnosed at birth.

Mr. Ian Stedman: It took almost two years.

The Chair: She probably had this at birth, though.

Mr. Ian Stedman: Yes.

The Chair: You did, too, probably.

Mr. Ian Stedman: I've had it my whole life.

The Chair: Did your parents?

Mr. Ian Stedman: My mother was also diagnosed. I just keep her out of it.

The Chair: Your mother was diagnosed after you were diagnosed.

Mr. Ian Stedman: Yes, in her sixties.

The Chair: Where does this come from?

Mr. Ian Stedman: It's genetic. We don't know. We can't trace it back. Her mother was already on her deathbed. We don't know where it came from after that. We've since found it in two of her siblings and some of her nieces and nephews.

The Chair: Imagine that. Where did the Muckle-Wells syndrome come from?

Mr. Ian Stedman: When we were diagnosed, we were numbers 11, 12 and 13 in Canada, but there's an umbrella, so there is a larger disease that we are one of the sub-variants of. We thought at the time that there were about 40 people in Canada with it. Now we're in the hundreds; we're identifying a ton. Hopefully, we'll be able to figure that out, but there's no knowledge.... It's been identified and treated, but no one really knows the etiology of it.

Dr. Alex MacKenzie: Often, we doctors who identify it very generously name it after ourselves. It's part of our altruism.

Voices: Oh, oh!

The Chair: Thank you very much, everyone.

Thank you very much, committee members and our analysts and our clerk, for your good work.

The meeting is adjourned.