EXPERIENCES OF RARE DISEASE PATIENTS

CANADIAN ORGANIZATION FOR RARE DISORDERS

Full report

February, 2023

CORD Canadian Organization for Rare Disorders

Ipsos

No one has a better understanding of what it means to have a rare disease than the patients who are affected by them. The input of these patient volunteers is absolutely invaluable, and our government has committed to making their experiences and perspectives an important addition to our scientific approach to drug assessments for rare diseases.

Honourable Rona Ambrose, 2014 Government of Canada

Contents

Study Objectives

Study conducted among patients and caregivers to help inform Canada's new Rare Disease Strategy, on behalf of The Canadian Organization for Rare Disorders (CORD).

OBJECTIVES

1. To understand current perceptions and experiences around navigating and accessing health care for patients with rare diseases and their caregivers
2. To give voice to patients with rare diseases and their caregivers in shaping policy to improve access to care and drugs related to rare diseases.

This report includes the results from two phases of research among patients and caregivers:

- **Phase 1**: an open link online survey distributed to CORD's database
- **Phase 2**: one-on-one face-to-face interviews with patients and caregivers

Canadian Organization for Rare Disorders

Phase 1: Online Survey

WHO WAS INTERVIEWED?

- Canadian Organization for Rare Disorders (CORD) emailed a 15-minute open link survey to its database, offered in English and French
- The link was also shared at the Rare Disease 2022 Fall Conference, November 21-22, 2022
- In total, 528 respondents completed the survey

FIELDWORK DATES

- October 26 to November 28, 2022

DATA WEIGHTING AND CREDIBILITY

- By the nature of this sample, the data was not weighted
- A margin of error cannot be calculated when utilizing a convenience sampling approach (open link) and the population universe is unknown
- Convenience samples are at risk for both sampling bias and selection bias
- However, our approach allowed us to access a large, valid sample size of patients with rare diseases/caregivers that would have otherwise been very challenging to reach

Phase 2: In-Depth Interviews

WHO WAS INTERVIEWED?

- In total, 10 virtual interviews were scheduled and completed
- Profile of respondents reflected patients by:
  - Province / Region
  - Gender
  - Caregiver / Person living with rare disease status

FIELDWORK DATES

- January 25 to February 10, 2023

[For the profile of survey respondents and interview participants, please click here to go to the Demographics section]

EXECUTIVE SUMMARY

Diagnosis Journey

Long path to accurate diagnosis for many, with multiple HCPs and possible misdiagnoses

Many patients living with rare disease are often waiting years to receive a final, accurate diagnosis. For one-third of respondents, it took 3 or more years for a diagnosis, and 15% saying it took 1 to 2 years. Half state it took less than a year and the average time among respondents is 3.7 years. The average number of healthcare professionals seen between the time of first symptoms to final diagnosis was 5.7. Additionally, six in 10 received an incorrect diagnosis. The average number of inaccurate diagnoses is 3.2.

Many of those interviewed spoke of how diagnostic delays and incorrect diagnoses take their toll on the mental health and well-being of patients and their caregivers. With delays in accurate diagnosis, patients also see their illnesses progress and potentially increase in severity.

Perceptions that healthcare professionals have limited awareness of rare diseases

A majority of respondents do not think their GPs (80%) or pediatricians (62%) are aware or informed about rare diseases. Further, almost half do not believe specialists have up-to-date knowledge and expertise about rare diseases. In some cases, interviewees spoke of the indifference, dismissiveness or even callousness from their physicians who are not familiar with rare diseases.

> *…It is true that great harm is being done to many because of* ***lack of awareness and even, sadly, callous indifference.***

Need for greater continuity of care for patients with complex rare diseases

Importance of continuity of care and the role of a general practitioner

For patients with rare diseases, there is a need for relational and management continuity of care. Those without a GP to oversee their health report missing important milestones in their care. This can include late referrals to specialists, missing lab results, delayed diagnosis and uncoordinated continuum of care.

Informational continuity of care is also critical for patients with rare diseases as they tend to see many HCPs since the start of their symptoms. Transferrable EMRs can play a role in the provision of informational continuity but many systems are not connected. This leaves patients needing to tell their story over and over again.

> *All the specialists just disappear, they all walk in and walk out. I'm convinced that **the medical system has become so siloed with specialists that nobody takes over overall responsibility**, and nobody considers the family and wider background.*

Needing to self-advocate

Patients and caregivers are often forced to navigate the system on their own, which leads to many having to self-advocate for their care. This requires them to look up more information about their disease on their own and motivates them to do more research and reach out to patient groups and supports. Further, participants also felt the need to advocate for other support services, such as mental health care. The needs of living with a rare disease go far beyond access to medications.

> ***Everything's on your dime. Everything's on your initiative and your efforts**. Heartbreaking efforts to say we matter. We care. We're suffering. And I don't say this feeling sorry for myself, and I'm well aware that the care homes are full of people who are suffering, and they all deserve enough compassion for, however, **for rare diseases, you are absolutely on your own.***

Need for more information about patient supports and education for community services

Gap in patient support information and counselling

There is a need for more consistent information provided for patient supports. With online resources now easy to access, a majority of patients/caregivers feel they know where to get information to deal with their disease, and only six in 10 say they have access to needed info at this time.

However, only 4 in 10 say they were provided with information on patient supports, e.g. contact person or number for additional information, or patient organization of support group. Patients and caregivers are left with the need to do personal research about their disease. There is a gap in the support and guidance that is made available to them.

There is also a lack of counselling support for patients with rare diseases. Seven in 10 did not have counselling support at the time of diagnosis or did not receive appropriate emotional and psychosocial support.

Community services

There remains a need to raise awareness and inform community services including educational, disability and employment services. Patients can face stigma and bias if support services staff lack awareness of issues around rare diseases. Less than one in 10 agree that educational services, disability services and employment services are aware and informed about rare diseases.

Patients can feel lost and in need of connection with others with rare diseases.

Only **58%** feel they have access to all of the appropriate information they need at this time

Only **43%** were given a contact person/number for additional info

Only **38%** were given information about patient organizations or support groups

Treatment and burden of incurred costs

Access to treatment and appropriate services

Only six in 10 have access to appropriate drugs for their rare disease. Roughly four in 10 are unable to access approved medicines and half say it is difficult or stressful to access medicines. Over one-third say costs for medicines are prohibitive.

There also remains a significant proportion who are lacking access to services. The largest gaps in access to care include access to specialists, clinics, care coordinators and counseling support. Furthermore, 7 in 10 do no have access to appropriate emotional and psychosocial support, a critical area of support needed for mental health wellbeing.

Six in 10 say they have incurred costs related to care and plan(s) used to cover drug costs. Other costs include daily expenses (parking fees) and the time lost to missing work and being at medical appointments.

Participants also describe a discrepancy of drug availability in Canada, between provinces. Respondents have learned that availability of treatment and standards of care also differ across countries (US, UK and Australia), and some have even considered moving elsewhere for their treatment.

Issues around lack of access to appropriate drugs

Only 60% have access to appropriate drugs

43% cannot easily access approved medicines prescribed (68% aware of Health Canada-approved medicines)

49% agree it is difficult or stressful to access medicines for my RD

36% are unable to access prescribed medicines because of costs or co-pay is too high

63% **pay out-of-pocket expenses**

Average spend	Median spend
$11,975.40	**$3,464.00**

a year on health-related expenses including prescription and non-prescription drugs, support services, health services, medical equipment and other expenditures.

Over the course of a lifetime, on average, this could add up to between half a million to a million dollars.

The majority selected rare disease centres as the model of choice for the national disease strategy when asked to choose between this and a national list of drugs.

84% support federal oversight (provide funding, support and standards for a coordinated inter-provincial rare disease strategy) to improve access to treatment and care for those living with rare diseases.

The main reasons are "more equal access across Canada" and "sets of standards/consistent policies."

Should the provinces maintain oversight, and each establishes its own rare disease strategy, patients and caregivers lean towards **setting pre-conditions tied to federal funding.**

*There is a need for this kind of centre for **access for ALL Canadians** regardless of the population of a province. Currently people in provinces with small populations have to travel to Ontario for treatment and diagnosis. **We can do better**. Rare diseases are treatable and a national strategy for a huge landmass with just over 40 million people makes sense.*

*A centre with specialists of different backgrounds working on the same problem **opens doors for collaboration and optimal results**.*

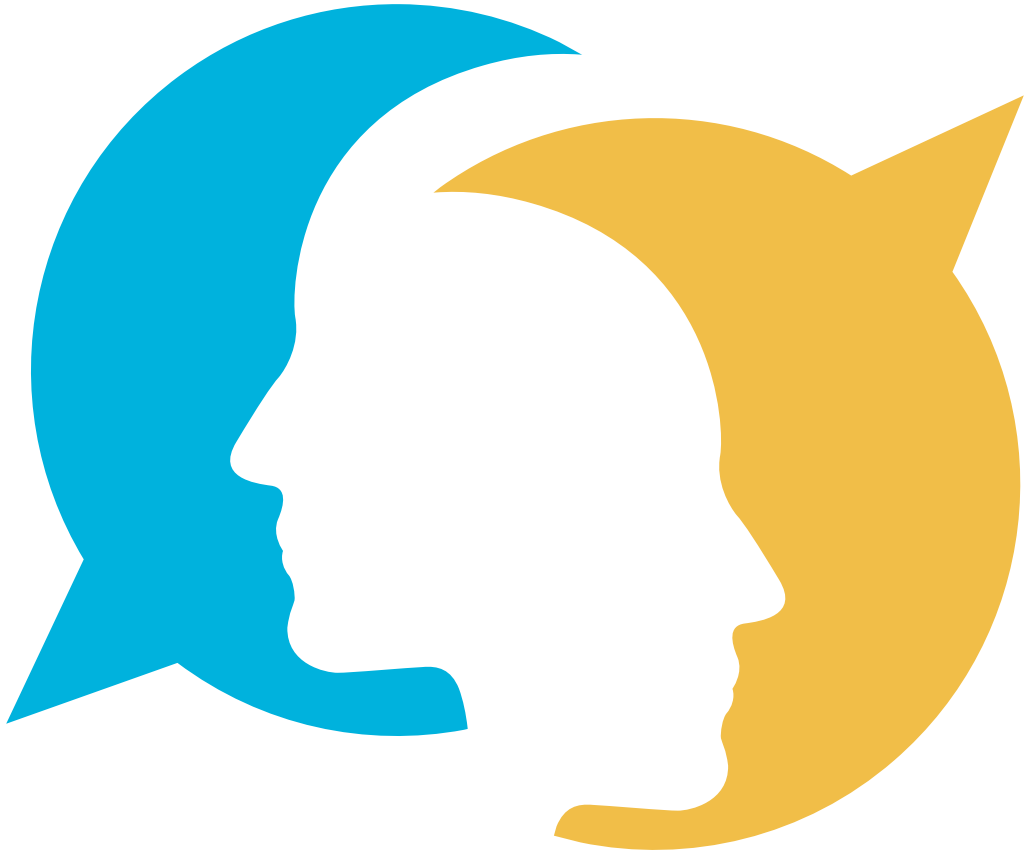
*We need health models that **are uniform across Canada**, and because provincial politics are so subject to ideological influences and the imperatives of getting elected. I think most of all that funding and commitment has to be well supported and long-term. I also think **a national body has a better chance of attracting the brightest and most committed** to making lasting change.*

From the voices of patients and caregivers

Responses to the National Rare Disease Strategy

POSITIVE OUTLOOK FOR HOPEFUL CHANGE

- Opportunity to bring more awareness, education and interest to rare disease of all kinds
- Focus of research that will drive innovative advancements
- Strength in numbers, centres will bring people and their stories together
- Will cost the healthcare system less money in the long run as more money and attention is allocated to diagnosis, monitoring and prevention of further adverse events
- Centralized approach will help mend our fragmented healthcare system



ALTHOUGH SOME SKEPTICISM AND CONCERNS REMAIN

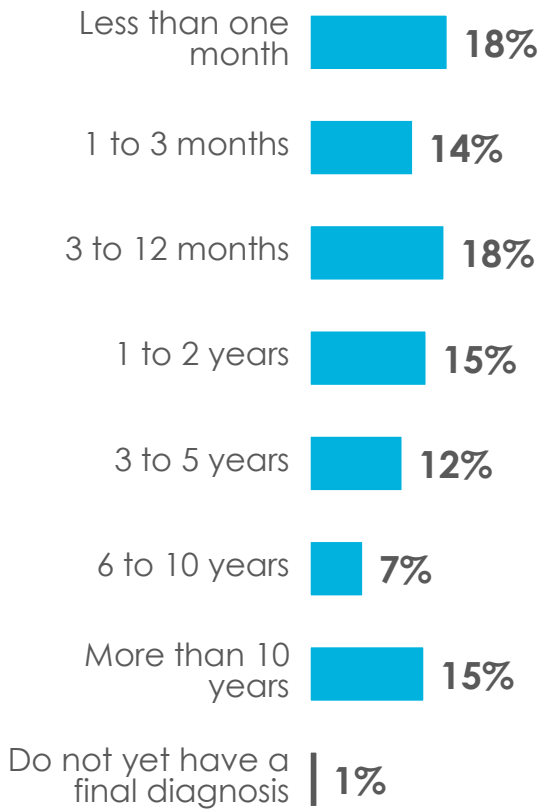
- Less talk, more action
- Need for equitable delivery across provinces so everyone can benefit
- Specialists are already limited' are there enough staff to properly run these centres of expertise?

DETAILED FINDINGS

DIAGNOSIS OF THE DISEASE

Challenges to timely and accurate diagnosis

LENGTH OF TIME TO GET ACCURATE DIAGNOSIS FOLLOWING FIRST SYMPTOMS OR SUSPICION OF DISEASE



AVERAGE
3.7 YEARS
TIME TO ACCURATE DIAGNOSIS

MEDIAN
1.3 YEARS
TIME TO ACCURATE DIAGNOSIS

Q6. How long after the first symptoms or suspicion of disease did it take to get an accurate diagnosis?
Base: All respondents answering (n=510)

Many patients with rare diseases are often waiting years till they receive a final, accurate diagnosis, with one-third of respondents saying it took 3 or more years, a further 15% saying it took 1 to 2 years, and one-half saying it took less than a year. The average time to correct diagnosis is 3.7 years.

A primary barrier to timely diagnosis is a lack of familiarity with rare diseases among doctors and a lack of recognition of symptoms.

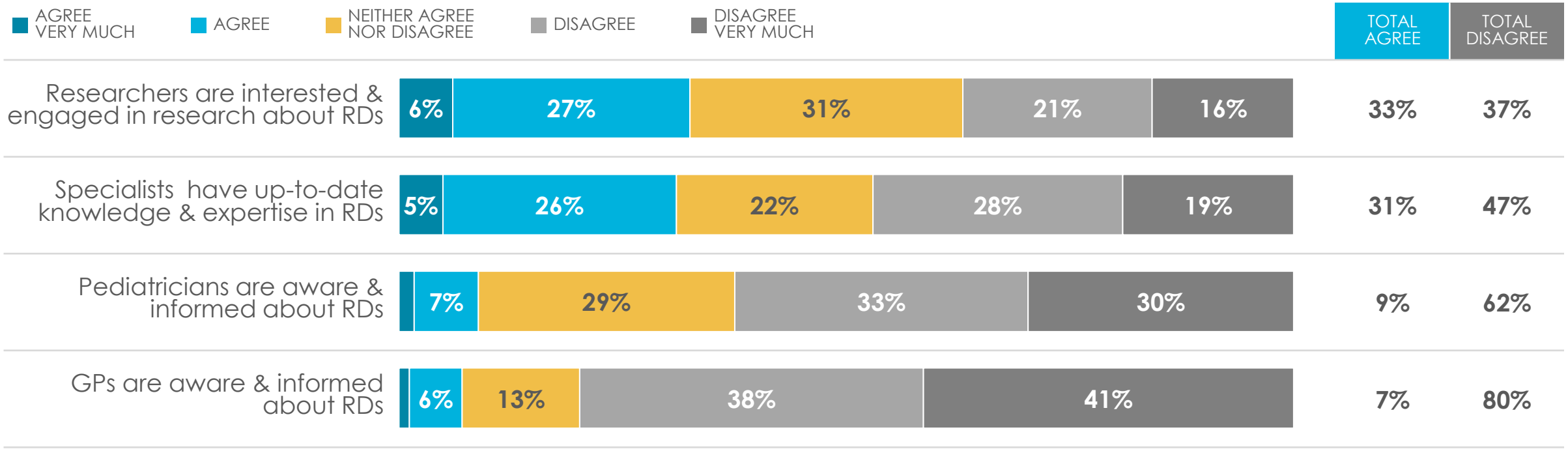
*Yeah, it was a long journey. It is rare disease, **so it's challenging and misleading at times**. It took **probably one year to get it right.***

*Doctors are not well versed in {rare} diseases and so you can get **people that don't get diagnosed or if they get diagnosed it gets changed** even by their existing doctor who decides he's read or talked to somebody else and said, well, I don't think you've got that, I think you got this. It's a serious issue in rare diseases…*

*Kind of looking back, I felt like something might have been changing in my body. I kind of feel like the pregnancy and that doctor was sort of a **blessing in disguise because I could have gone years without diagnosis**. I would have brushed {the symptoms} off. Like personally, I would have put it off. I would have thought, {its an effect since} I changed my diet or I should get on the treadmill… I can see sort of a combination of maybe personal {doubt} along with a strained healthcare system, how people can go for a couple years, undiagnosed.*

Very few agree that pediatricians and GPs are aware and informed about rare disease. Even half perceive specialists as lacking in up-to-date knowledge and expertise.

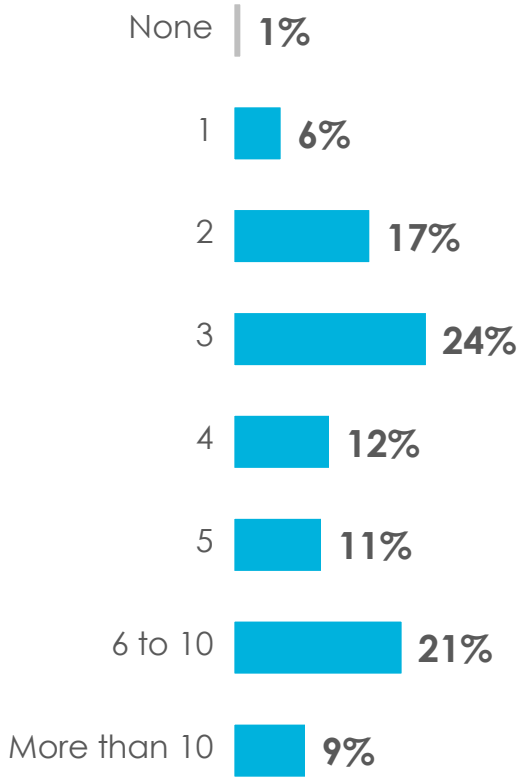
*About people with rare disease, **they spend very little education on it**. When I attend conferences and ask physicians how they are, how they're educating younger physicians on rare disease, there's no good answer.*



Q10. To what extent do you agree/disagree with the following statements about information about rare diseases overall?
Base: All respondents answering (n=510, 293, 487, 473) Note: <3% are not labeled, statement have been shortened, full-text may be found in the appendix

Majority of patients see at least three HCPs before final diagnosis

NUMBER OF HEALTHCARE PROFESSIONALS SEEN BETWEEN TIME OF FIRST SYMPTOMS AND FINAL DIAGNOSIS



AVERAGE
5.9
HEALTHCARE PROFESSIONALS SEEN
(EXCLUDING NONE)

MEDIAN
3.2
HEALTHCARE PROFESSIONALS SEEN
(EXCLUDING NONE)

On average, patients see 5.9 healthcare professionals before final diagnosis. Patients mentioned how a lack of awareness of rare diseases can also lead to doctors being dismissive and patients needing to consult many professionals.

*…It is true that great harm is being done to many because of **lack of awareness and even, sadly, callous indifference.***

*If one of my doctors decides to move away and I have to go find somebody else, I'm going to go through the same thing where you got doctors **who either don't believe in it or don't understand it or don't care.***

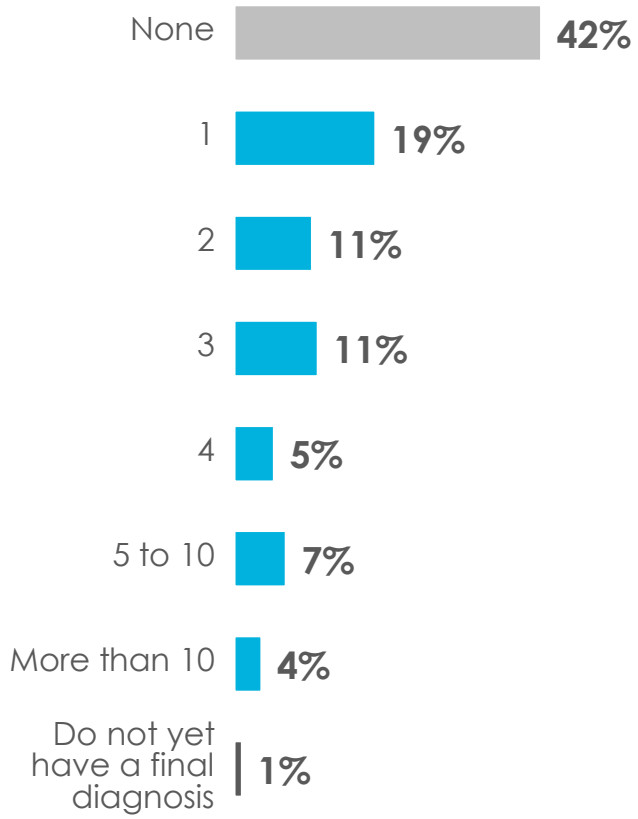
*Insinuating different things that were incorrect and was almost lecturing me and I thought, **OK, you're supposed to be my advocate. You're supposed to be helping me through this**, you know, you shouldn't be lecturing me, {you could be} doing this or you could be doing that. You know that?*

*I had one guy, he examined me and says, 'you know, we're not sure what it is, but we're going to check it out. I want you to come back in two months and I'll be on your case and don't you worry'. I said OK. I came back in two months; **he started lecturing me that I'm probably not really that sick** and you'll get better don't you worry. He asked why did you come anyways, and I said because you asked me to come. I'm thinking, oh goodness. I left that appointment actually pretty upset.*

Q7. How many health care practitioners (HCPs) did you see between the time of first manifestations or symptoms and the final diagnosis? If you have not yet received a final diagnosis, please indicate the total # HCPs consulted to date. This could include your family doctor, pediatrician, other medical specialists, physiotherapist, psychotherapist, or others.
Base: All respondents answering (n=521)

Six in 10 receive at least one incorrect diagnosis

NUMBER OF WRONG OR INACCURATE DIAGNOSES RECEIVED PRIOR TO THE FINAL DIAGNOSIS



AVERAGE
3.2
WRONG / INACCURATE DIAGNOSES
(EXCLUDING NONE)

MEDIAN
1.9
WRONG / INACCURATE DIAGNOSES
(EXCLUDING NONE)

Q8. How many "wrong" or inaccurate diagnoses did you receive before the final "right" diagnosis?
Base: All respondents answering (n=518)

Of these six in 10, the average number of wrong/incorrect diagnoses is 3.2. Incorrect diagnosis can steer patients to wrong treatment paths that could inadvertently harm the patient or negatively impact disease prognosis.

*Because **the physician that was treating me was convinced it was something else** and I was so convinced from the research that I had done and the symptoms that I was feeling. And that's something that I had to push for until they got the point that they're not going to get rid of me. So that was the kind of process it was. It was frustrating and it was always sly, very unnerving. I **don't know what the repercussions** of having that process, being that lengthy, really means at the end of the line.*

*I've read online there's a lot of issues with ophthalmologists, that there is a **tendency to minimize what they're seeing** because there's some neurologist that don't like to diagnose my {disease} because it's such a hard disease to treat.*

*The doctor had made a comment that in his 36 years of practice he had **never come across a patient** with {rare disease}. So, it kind of shows you how rare it is or how people go undiagnosed, possibly, and **it's chalked up to** asthma or COPD or **other issues that have the same relatable side effects or symptoms.***

*The other significant aspect is a misdiagnosis of a rare disorder or any disorder **results in inappropriate treatment**. We are very far from patient centered care. In fact, it almost doesn't exist.*

There is a sense of loneliness and emptiness and a tremendous toll on the mental health of patients with rare diseases and caregivers

People living with rare diseases have different lived experiences. They could be not working, living with disability and struggling with other health issues – including mental health.

*You had all these hopes and dreams and expectations that you could do with your child for your child, and it just kind of **all comes crashing down** and then on top of that cause it's a rare disease and nobody knows anything about it.*

*It's a very lonely island that you're sitting on because **people forget that you are people**. And everybody is willing to be **sympathetic but not empathetic**.*

*I don't spend money on my care because I gave up going to therapy, searching for treatment options a long time ago. I've been admitted to hospital a number of times, seen many GP's and specialists, so if you can quantify that, then you will have your answer as to how much we spend on treatment for one patient. However, this doesn't include **therapy for mental health treatment** which {with my rare disease} is extensive.*

*We've got denied because {disease} **isn't considered life and limb threatening which is total garbage**. Canada won't fund a rare disease drug, or Ontario I should say. It has to be life or limb threatening and what bothers me is that with {disease} comes very, very severe depression and suicidal tendencies, so a lot of our population are not working. I'm considered a high functioning patient, but most of the people I talk to don't work. They live on disability, they can't work because of their mental health status. And I think that's important to note because if you're talking about life and limb threatening, there is that **mental health aspect that I don't think the Government of Canada considers**.*

Continuity of care is critical, especially among patients living with rare diseases

Some interviewees identified a need for relational and management continuity of care with a healthcare provider to oversee medical and mental health needs, especially those without a family physician. **Patients who lacked a GP missed out on important milestones in their disease progression, including late referrals, missing lab results and delayed diagnosis due to the lack of someone central coordinating their care.**

All the specialists just disappear, they all walk in and walk out. I'm convinced that ***the medical system has become so siloed with specialists that nobody takes over overall responsibility****, and nobody considers the family and wider background.*

I think its probably a ***national problem finding a family physician****. Because I can't do anything. I, you know, I really had to plead my case and say, look, I really do need somebody that's going to manage this because I can't.*

I have a son who has a rare genetic disorder that was only identified probably about 10 years ago. For about 25 years he went with guesswork. For best intensive purposes, {different diagnosis} was the best diagnosis they could give so that we could get support. ***Just out of sheer luck we get a new family doctor who went through all his records*** *and said, hey, there's something, there's a file here from a geneticist. That {my son} has the key identifier for the {rare} syndrome, and it's in a note. I {family doctor} want to follow up with genetics... So we kind of lucked out.*

There is particular concern for the continuity of care from pediatrics to adult care for patients with rare diseases.

{My child} is five to six years away from transitioning to adult services. ***I'm actually quite terrified for the transition*** *because there isn't the same {level of services}, like the there is a huge shift in the quality of service that you get from pediatrics to adult care.*

Informational continuity is also important for patients with rare diseases who see multiple healthcare providers. Patients are carrying the burden of managing their personal records. Fully connected EMR systems are still lacking in Canada.

One of the other things we've learned from people in the US, and we're noticing it more and more in Canada now is they ***keep impeccable records****. So if they go to the doctors today, they have a record of that. I don't know if they pay for it or I don't know how it works, but we've met people and they come to the conferences and* ***they have files like this thick****.*

When {my son} first moved from pediatric care to an adult hospital…they wanted they wanted to know when he had his last surgeries and he's like "Oh my God, I don't remember like don't you guys have that records? No, they didn't transfer them to me." So, I had to drive from {adult hospital} to {the children's hospital}. It's not that far. Pay $40 to get my records and then bring them back to the to the {adult hospital} and say here take whatever you want. I don't care. But you know, and there's that thing we learned from the Americans that we've met, you know, ***keep all your records****… Now {we have} all that so it it's really good.*

Despite stories of negative experiences, a few patients also shared some positive experiences and expressed pride in Canada's public healthcare system.

He did the most marvelous act, I handed him a note with my wife's symptoms… he stood up and he said I am going to get you some help. And he walked around and put his hand on my shoulder. That single act of grace has sustained me for this horrifying ordeal. And that will stay with me all my days. That is the ***healing touch. The human connection****.*

The only reason I was diagnosed is because I went to a physician in Toronto - he's the specialist who diagnosed me and it's ***only because he had one other patient*** *with {the same disease} that he was treating.*

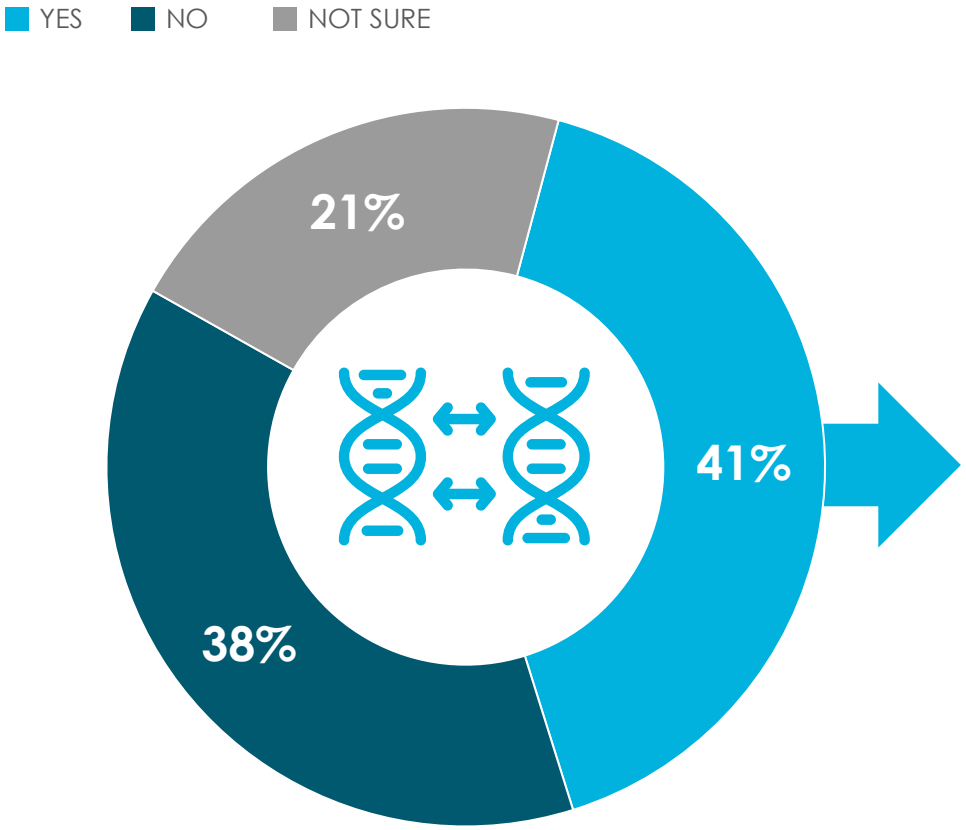
No, we're not moving from here right now, like ***we are absolutely lucky with the healthcare we have.*** *So, you know, when we hear people that complain about healthcare, we go well, knock on wood, we're super lucky with what we have.*

We have a really good team. And I would ***say it's like a bingo card. I'm pretty sure we've collected every square****, every possibility. But they're great and they work very well together.*

GENETIC DISEASES

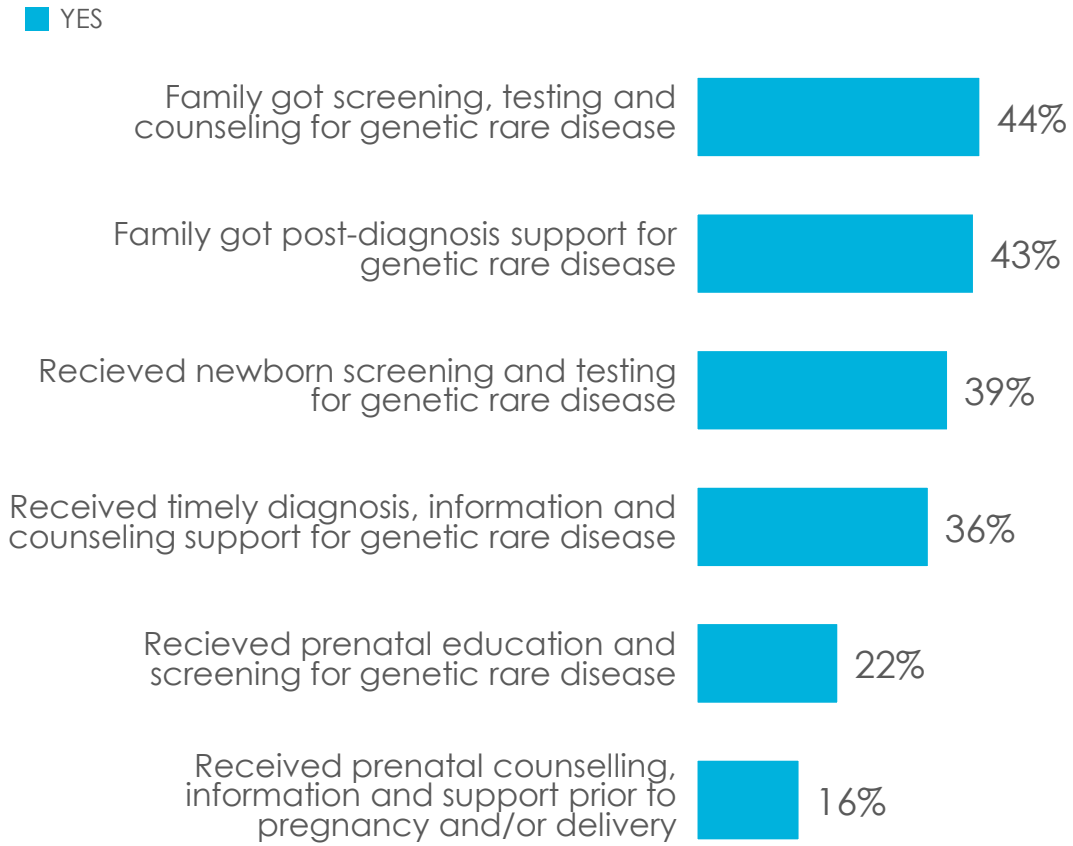
Rare Disease Genetic or Inherited Condition

GENETIC OR INHERITED CONDITION



Q16. Is the (your) rare disease a genetic/or inherited condition?
Base: All respondents answering (n=526)

GENETIC SERVICES FOR RARE DISEASES – AMONG "YES"



Q17. For the following statements about genetic services for rare diseases …
Base: All respondents answering (n=128, 123, 133, 139, 184, 181)
Note: statement have been shortened, full-text may be found in the appendix or questionnaire

Management of Genetic or Inherited Disease

Jump through many hoops for genetic testing – even resorting to private companies

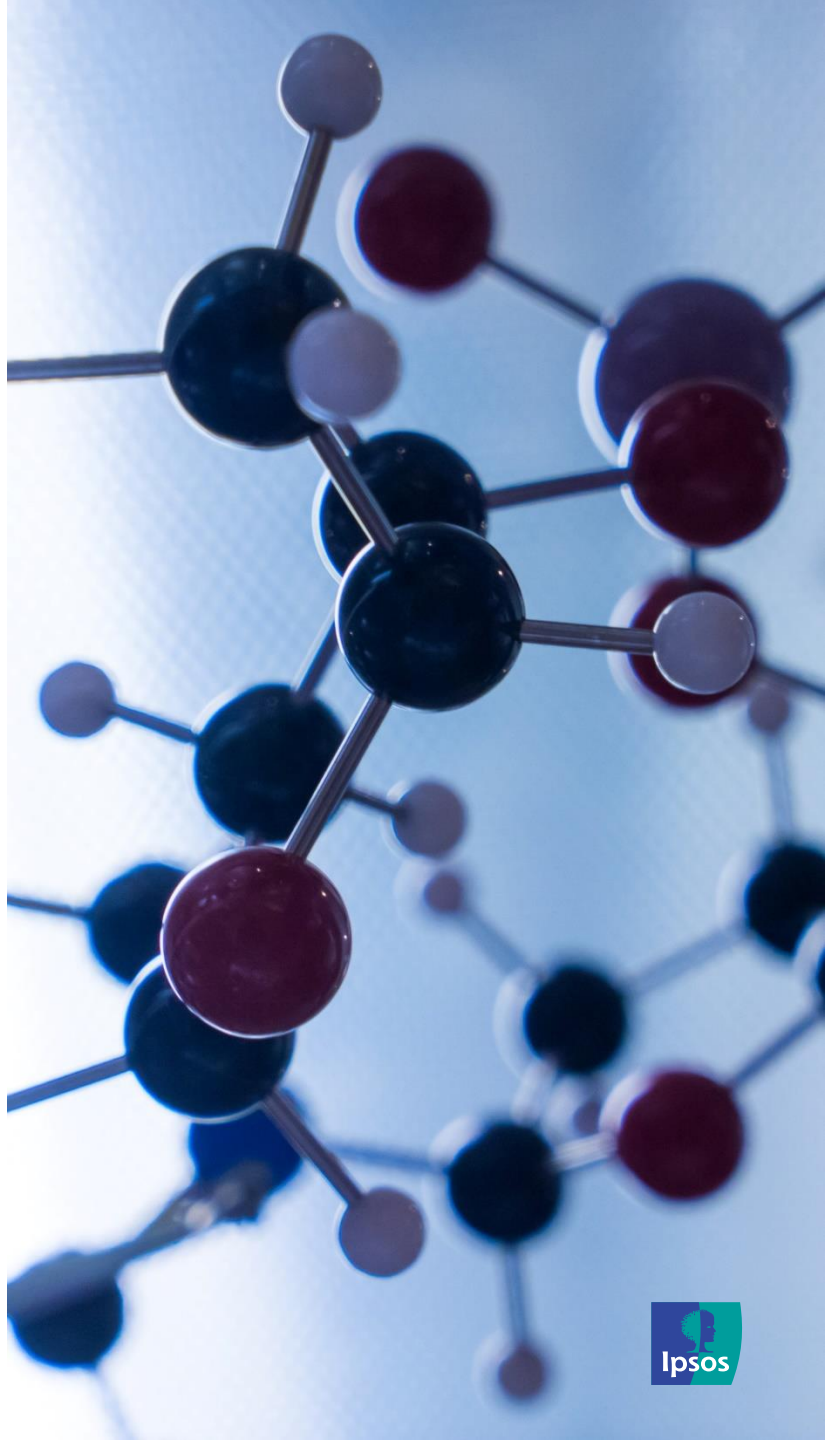
*There is a shocking and evident lack of education among doctors, nurses and care aides at all levels - that is in my experience. This is **especially true of rare diseases and genetic factors**, including at the most practical level. There is also rampant cynicism, indifference and even a callous manner in responding to patients or their advocates. **Contrary to the professed lofty goals a lot of harm is being done**. My wife and I have experienced the lot.*

***No one did any testing. No one did any traceability**. I did do genetic tests only when I participated in a clinical trial in the US.*

*I had to go to {a USA university} and all my own efforts to even begin to approach this and get some help. I say that mindful of the fact that there are some wonderful helping people, but my God, the **people in the commercial industry have been the most help to me**, exceptions such as UBC neurology department has been wonderful.*

More considerations are needed in diagnosis, especially records of family history

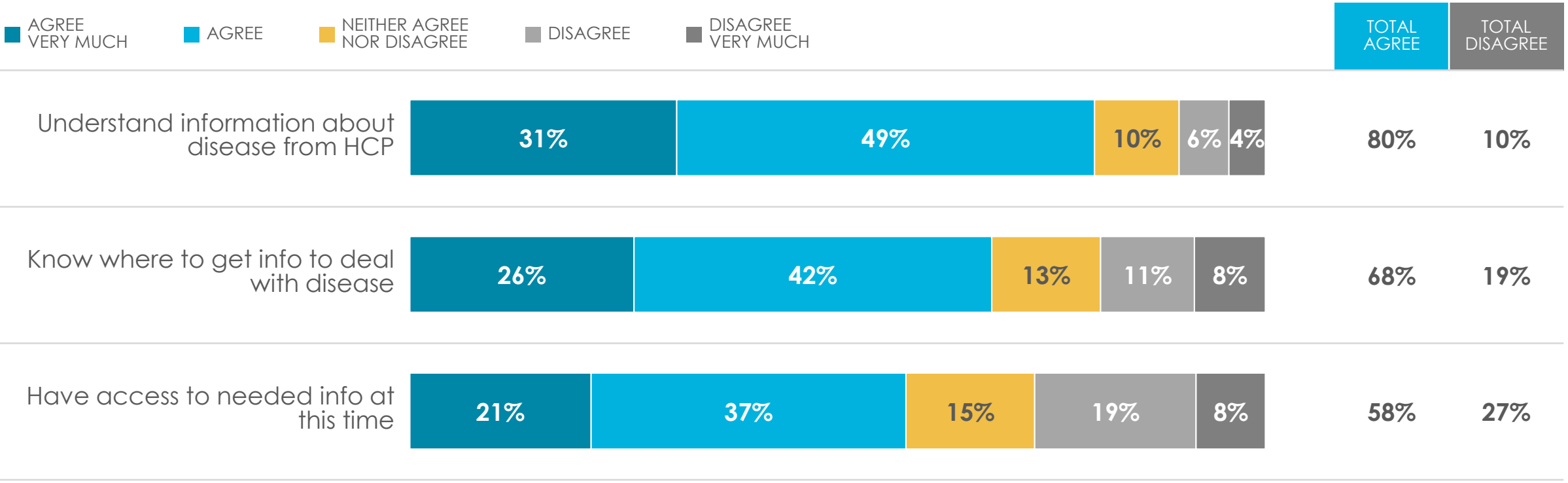
*I did speak to the neurologist and her family history was described. Deep, remarkable, medical history. **No one really took that into account** until much later.*



ACCESS TO INFORMATION AND SUPPORTS

Small but significant proportions of patients say they don't understand the information provided to them from their HCP about their disease, or don't know where to get info to deal with the disease. One quarter say they don't have access to needed info at this time.

You can't even say. Well, now, what do we do? Because nobody can give you the answer. ***You're flying blind****.*



Q9. To what extent do you agree/disagree with the following statements about the information you have received about the rare disease?
Base: All respondents answering (n=516, 492, 503)

There is a lack of Canadian resources for patients to turn to

Those who sought out more information about their rare disease found more resources across the pond. There are more extensive support groups, advocates and patient information sites elsewhere. Canada is perhaps limited with the smaller population.

*But I can tell you **I didn't find the level of information and the level of up-to-date information on the Canadian website** {compared to UK}.*

*But it's like you leave there {the first appointment with a diagnosis}. **Somebody tells you have a lifelong life-threatening disease with zero information**. Lack of guidance, lack of like new patient info.*

***Credible is the keyword** there because I could look up anything online, I could put anything in Google. 1000 things pop up, but they all look legit. How do I know that's actually something that I should be following or finding out more about? So yeah, having a credible resource. Umm, I think is key there too.*

*No, nobody heard of it. There's a lot of people that still don't know about it in the hospital system. Moving forward, It's nice that there is the availability {of resources now}, but **back then there was nothing. It was really hard to find anything** on {my disease}.*

*There was no real one spot to go and find the information that could help me as a chronically ill person. And I just had to stumble upon it on my own. And I feel like there should be a place, **there should be a list somewhere** or something like that. Hopefully they would have a list of treatments available and a list of programs that you can go to and support groups, maybe, and just all of this stuff. Its kind of like **a one stop shop** for someone living with chronic illness. That is great, that sounds wonderful.*

Many patients self-educate over time and must become their own advocates

__Everything's on your dime. Everything's on your initiative and your efforts__. Heartbreaking efforts to say we matter. We care. We're suffering. And I don't say this feeling sorry for myself, and I'm well aware that the care homes are full of people who are suffering, and they all deserve enough compassion for, however, __for rare diseases, you are absolutely on your own.__

Physicians don't know how to order it. __I recently had to facilitate finding through my contacts__ who had the patient support program, how they could get drug and then get it out to a patient in BC, a friend of mine who needed it.

I find myself __debating my treatment__… I see what is done in other jurisdictions and I, you know, sometimes I question myself as to __why things are being done differently here__… I don't feel like I'm educating, but I'm often debating because I don't necessarily agree with what they're suggesting.

There's a lot of tiny {details}, __you do a lot of personal research__, and you find out a lot of things because what works for you doesn't necessarily work for somebody else with {the disease} it's very personalized.

I've been very blessed to have a very good GP that is quarterbacking it the way. I think a lot of challenges people have is if they don't have a good GP that's going to quarterback the disease and get the right specialist in and if that one doesn't work, get somebody else. They don't have that. __They're fighting the battles themselves__.

Caregivers play an integral role in the whole patient journey, although some feel barriers in acting as a patient advocate

Self-advocacy is important in navigating the current healthcare system. When patients are not able, caregivers' step in and play the role of the patient's main advocate.

There is a sense of community and bond among caregivers, who are facing similar hardships caring for a loved one living with a rare disease.

*Issues of being a guardian and a caregiver, but also a **legal** spokesperson, with an advanced care directive. On the moral grounds of autonomy.*

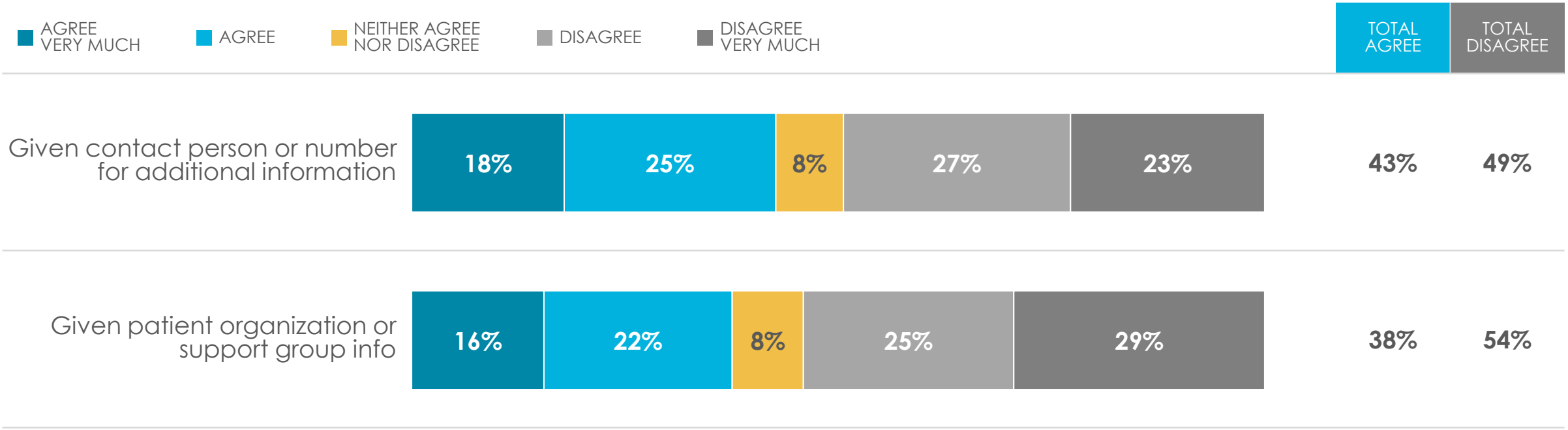
*{Advocate} that I get proper treatment and get treated properly by doctors and so I'm not shy to say my concerns. I'm not confrontational, but if I don't feel right, I'll say something. I may not say it in meeting, but I'll do it by going to a different doctor. But I think anybody who's got **any sort of rare disease has to really be their own advocate** or if it's a child or anything like that then the **parent has to do for that child.***

***Who has most interest in the disease, when I speak to them, are the caregivers**, the professionals, the higher you go, the less interest there is in you and less interest in your disease.*

*I am devoted to my wife's care so that she receives compassionate and appropriate care. I am also committed to helping research to not being angry about our experience and to help in bring about change in our knowledge and support of those suffering from rare disease. **Their families and loved ones also suffer**. **It is true that great harm is being done to many** because of lack of awareness and even, sadly, callous indifference.*

While patients with rare diseases could benefit from greater support, roughly half say they were not given a contact number of a person or patient organization/support group.

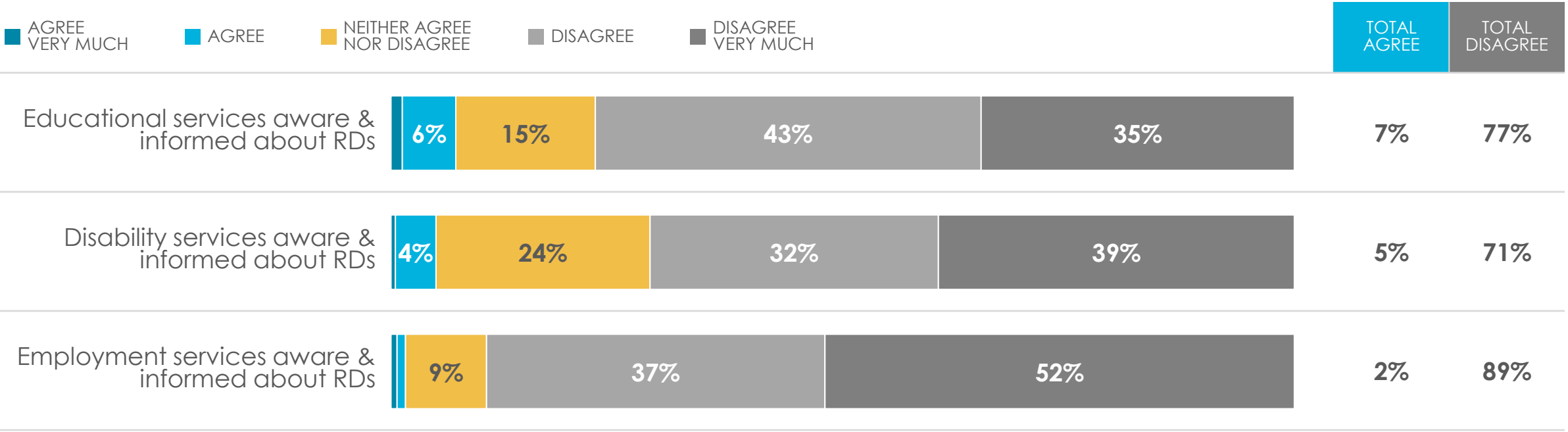
*It was rather **difficult to find any information {regarding my disease and supports} because I think I was probably the first person** in like in my city to have it. So they basically studied me, they researched on me. I've had people come in and say ohh I had to write a paper about you. You're the one that has {this disease}.*



Q9. To what extent do you agree/disagree with the following statements about the information you have received about the rare disease?
Base: All respondents answering (n=519, 519) Note: <3% are not labeled, statements have been shortened, full-text may be found in the appendix

Very few feel that community services are educated and familiar with the impacts of having a rare disease but some personal stories show some light

*Her **school, her teachers and the principals and the vice principals have been absolutely amazing** in talking with us and handling her, in accommodating her. They are fantastic and have been extremely supportive to her with at all and they help us. Because there's nothing here that talks about rare disease day, it's pretty much nonexistent, except for the people that we talk to about it. And the school has allowed us to come in and present and do an activity with the classrooms every disease day to try and make everybody aware. So her immediate peers all understand her disease and they all understand this rare disease day and kind of what it's about.*

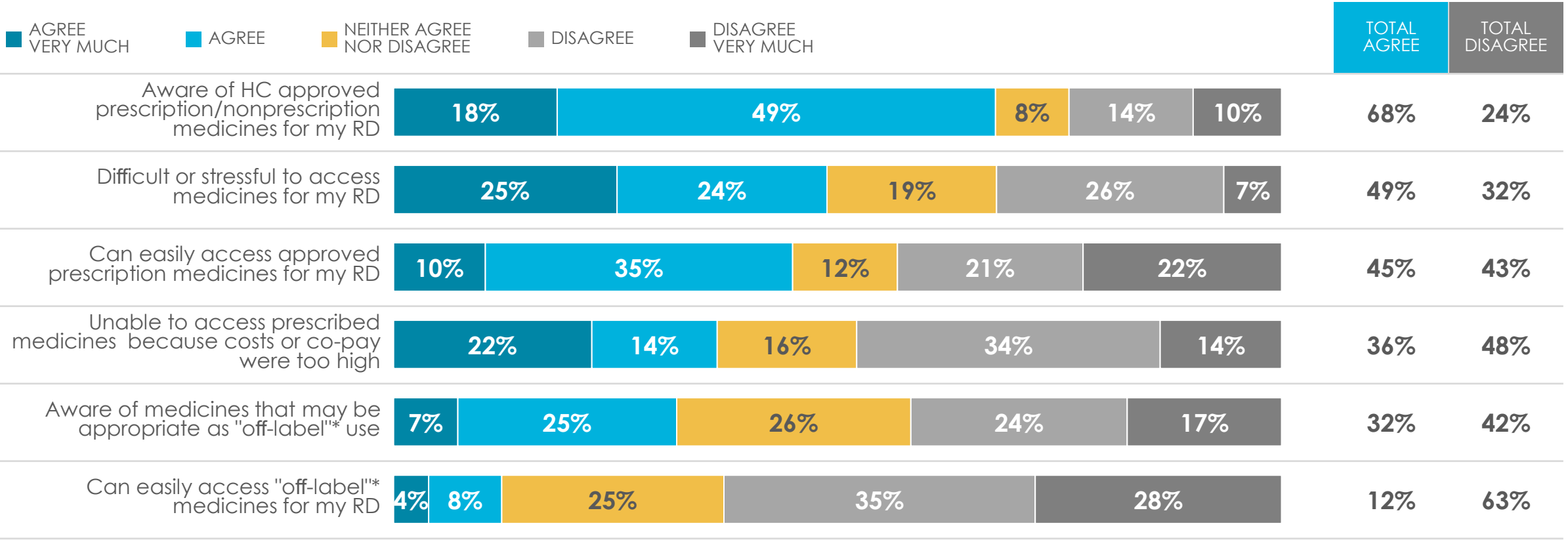


Q10. To what extent do you agree/disagree with the following statements about information about rare diseases overall?
Base: All respondents answering (n= 449, 435, 401) Note: <3% are not labeled, statement have been shortened, full-text may be found in the appendix

ACCESS TO CARE AND TREATMENT

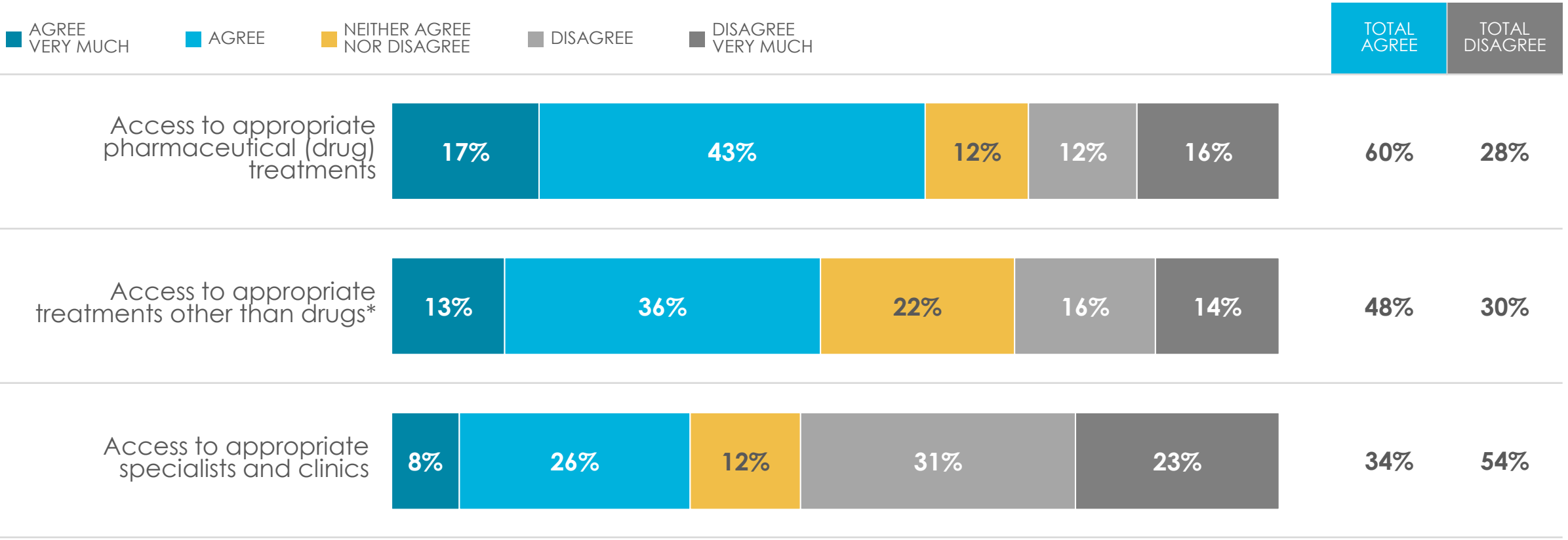
While almost 7 in 10 are aware of Health Canada's approved drugs for their rare diseases, fewer than half say they can easily access these approved prescription medicines.

- Over one-third are restricted by costs or co-pay of medicines being too high.
- Fewer, one-third, are aware of off-label medicines for their rare diseases, and only one in 10 can easily access these.



Q12. To what extent do you agree/disagree with the following statements about medicines for your rare disease? If your treatment includes more than one medicine, please respond with respect to your overall experience.
* Medicines that have been approved in Canada but not for their specific rare disease. Base: All respondents answering (n=472, 474, 345, 328, 461, 439) Note: <3% are not labeled, statement have been shortened, full-text may be found in the appendix

About 3 in 10 disagree that they have access to appropriate pharmaceutical drug treatments and other types of treatments. Surprisingly, only one-third have access to appropriate specialists and clinics, while over half do not.



Q11. To what extent do you agree/disagree with the following statements about the access to treatment and support you have received for the rare disease?
* For example, surgery, radiation. Base: All respondents answering (n=499, 416, 523) Note: <3% are not labeled, statement have been shortened, full-text may be found in the appendix

Inconsistency of treatment and support across Canada

Participants describe discrepancy of drug availability and specialists in Canada, between provinces.

They are also aware of treatments available elsewhere but not in Canada, and if offered in Canada, it is never first: differences in pricing, standards of care, and availability of clinical trials for patients.

Furthermore, with different standards of care, some participants have even considered moving elsewhere for their treatment.

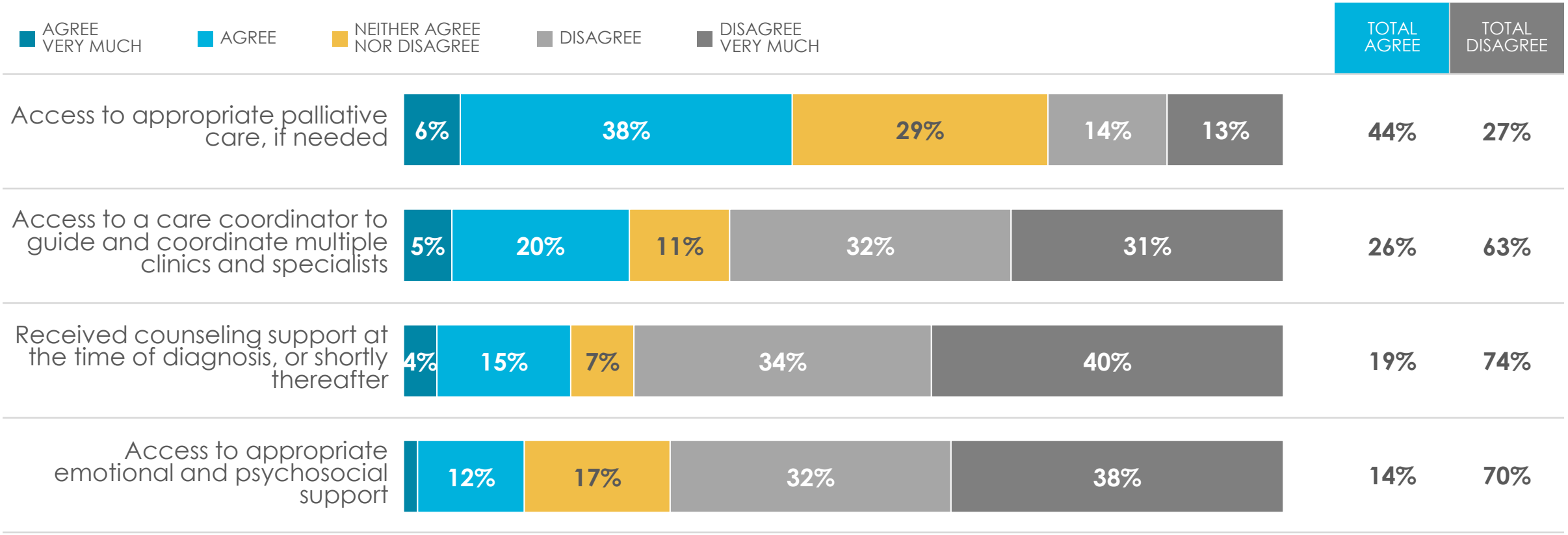
*Differences between province. That **unknown process kind of discourages me** from needing {treatment} unless I really feel like I need it. So right now I don't feel like I need it. I don't know if that makes sense, but I don't know. I don't even know if it's possible to be treated in an Ontario hospital.*

*I participated in the clinical trial phase two [US study with a trial in Canada]. And now we have no access to it [Phase 3 of the clinical trial was not run in Canada]. So you imagine someone who suffers with depression, getting a medication, **having the best summer of their life, and then taking that away**. And what it did to their mental health…. And I will be participating in {another clinical trial} in the US, but none of these companies have Canadian sites hardly. And when they actually launch, they launch in the US usually 1st and then in Europe and then Canada because {our} population is so small and it's such a rare disease... **It's really hard to get them to come to Canada.***

*I will be participating in {a clinical trial} in the US, but **none of these companies have Canadian sites** hardly. And then when they actually launch {the drug}, they launch in the US usually 1st and then in Europe and then Canada because our population is so small and it's such a rare disease.*

*The **whole island is really limited** for a lot of healthcare services. People have to go off island. And there's no real specialists or things here.*

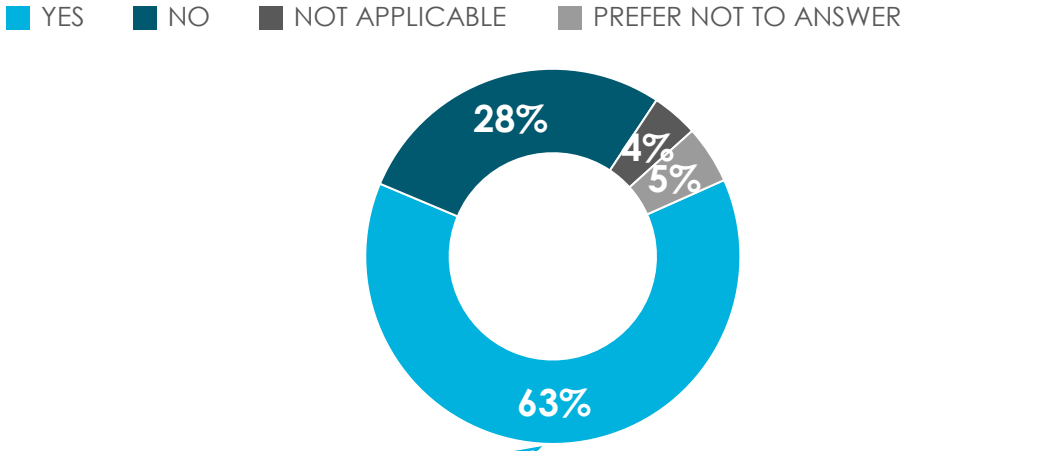
The largest gaps in access to care are with care coordinators (63% do not have access), counselling support at the time of diagnosis (74% do not have access), and further emotional and psychosocial support (70% do not have access).



Q11. To what extent do you agree/disagree with the following statements about the access to treatment and support you have received for the rare disease?
* For example, surgery, radiation. Base: All respondents answering (n=310, 491 500, 495) Note: <3% are not labeled, statement have been shortened, full-text may be found in the appendix

Two-thirds have incurred personal costs to care for rare disease, higher among those in SK/MB and lower in ON.

HAVE INCURRED PERSONAL COSTS TO CARE FOR RARE DISEASE



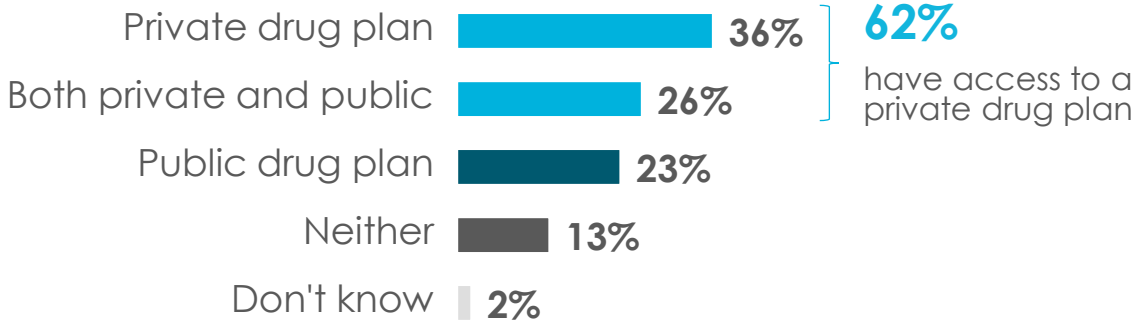
BC	AB	SK/MB	ON	QC	ATL
A	B	C	D	E	F
77% D	74%	**91%** DF	61%	**79%** D	63%

Q13. Do you and your family incur personal costs related to care for your rare disease, such as health services (physiotherapy, counselling, etc.), prescription or non-prescription drugs, support services or other costs?
Base: All respondents answering (n=526)

For some patients, costs incurred include daily expenses such parking, but also loss of time and missing work for medical appointments.

For others, the financial burden can be tremendous, with most costs focused on support services (average over $8K, and median is ~$2K) and prescription drugs (average over $7K, and median is ~$1K). The cost of health services and medical equipment is also significant (average over $2K, and median is ~$1K) with non-prescription drug costs following closely behind (average over $1K, and median is ~$500). Other expenditures varied greatly, with the range of $50 to $80K.

PLAN(S) USED TO COVER DRUG COSTS



Q15. Which plan(s) have you used to cover your drug costs for rare diseases?
Base: All respondents answering (n=526)

Annual Incurred Healthcare Costs to Care for Rare Disease

Among those who answered "Yes" to incurring personal costs for care	PROPORTION OF WHO PAY OUT OF POCKET	MINIMUM TO MAXIMUM RANGE (EXCLUDING "$0")		AVERAGE (EXCLUDING "$0")	MEDIAN (EXCLUDING "$0")
Support services *(education, childcare, elderly care, support for daily living, etc.)*	27%	$100	$100,000	$8,675.50	$2,350.00
Prescription drugs *(buy or co-pay)*	71%	$25	$500,000	$7,240.40	$913.60
Health services costs *(testing, physio-therapy, counselling, not including drugs)*	50%	$20	$15,000	$2,208.90	$975.00
Medical equipment *(wheelchair, electronic aids, monitoring devices, etc.)*	36%	$50	$20,000	$2,028.40	$852.90
Non-prescription drugs *(over-the-counter medications)*	59%	$30	$32,000	$1,005.00	$473.00
All other expenditures	50%	$50	$80,000	$3,234.90	$926.50
AVERAGE TOTAL		$26	$506,320	$11,975.40	$3,464.00

Q14. To the best of your knowledge, what is the amount your family spends annually related to care for your rare disease?
Base: All respondents answering (n=329, 324, 327, 325, 329, 327)

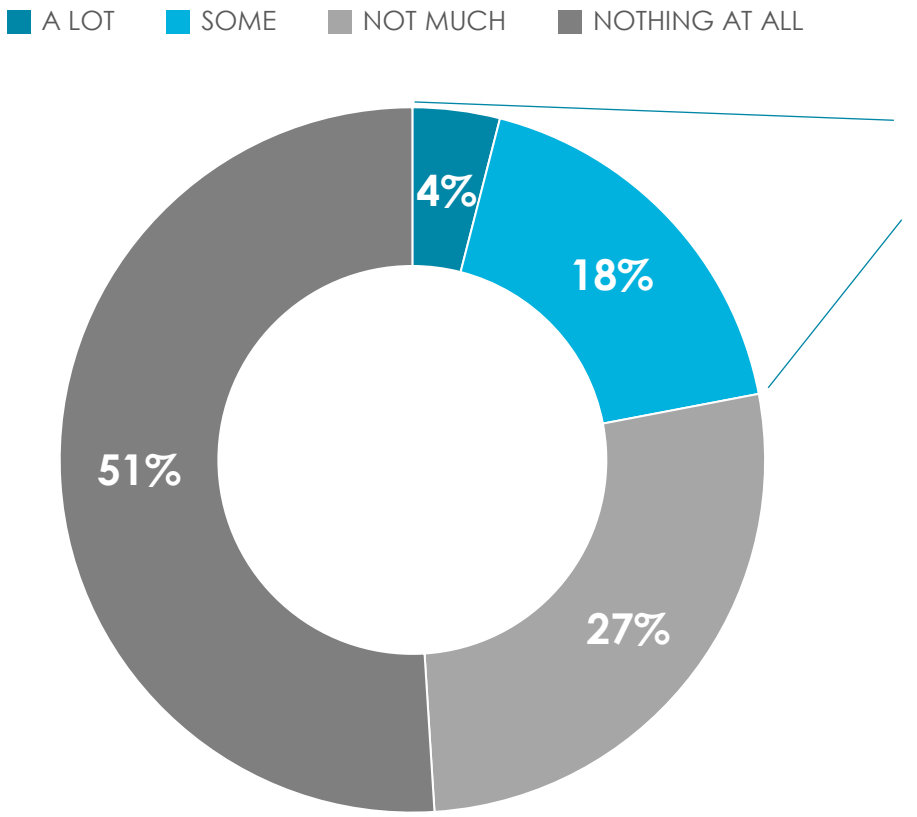
SHAPING THE NATIONAL RARE DISEASE STRATEGY

Awareness of a Canadian Rare Disease Strategy

Accessing drugs and treatments is challenging for many of those living with rare diseases in Canada. There are currently more than 7000 rare conditions that affect about 1 million Canadians. When medicines are introduced in Canada, **it takes much longer** to be made as part of government-administered drug programs. Further, clinical expertise and **understanding of rare diseases are limited**, highly specialized and **not consistent across the country**. At the moment, Canada **does not have** a national rare disease policy framework in place. Consequently, many clinical trials and treatments come to Canada later or do not come to Canada at all.

In the 2019 Budget, the Canadian government made a **commitment to work together with provinces, territories and other partners** to create a national strategy to improve timely and equitable access to drugs for rare diseases. This strategy, known as ***The Canadian Strategy for Drugs for Rare Diseases***, will include up to $1 billion over two years, starting in 2022–23, with up to $500 million per year, thereafter.

AWARENESS OF STRATEGY



22%

have heard "**a lot**" or "**some**" about *The Canadian Strategy for Drugs for Rare Diseases*

Q18. Before this survey, how much had you heard about the Canadian government's commitment to invest in a national strategy for drugs for rare diseases?
Base: All respondents answering (n=528)

Respondents in the survey were presented with the following descriptions of two potential models for *The National Rare Disease Strategy…*

Currently, the national rare disease treatment strategy is still **under development** as it prepares to launch on or before March 2023. As Canada considers how best to create a national strategy on drugs for rare diseases, it is important that **Canadians continue to have a voice** in helping to shape it.

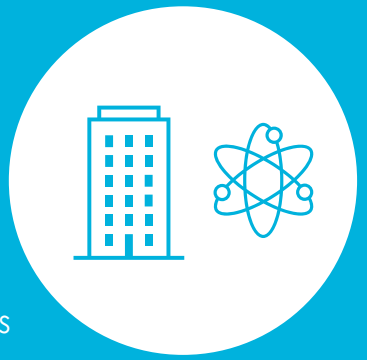
Very broadly, there are **two potential models** for the national rare disease strategy.

MODEL: RARE DISEASE CENTRES

Funding would be invested in **specialized rare disease centres** with clinical experts providing **care based on best practice guidelines**. The centres would offer screening and diagnosis, access to drugs, referrals to specialists, access to clinical trials, and research. Most of the **functions of proposed Rare Disease Centres are already in place** within the health system but **would be coordinated** and **appropriately funded**. Patients across Canada would be able to access the appropriate specialist centre, **regardless of where they live**.

In addition, these sites will:

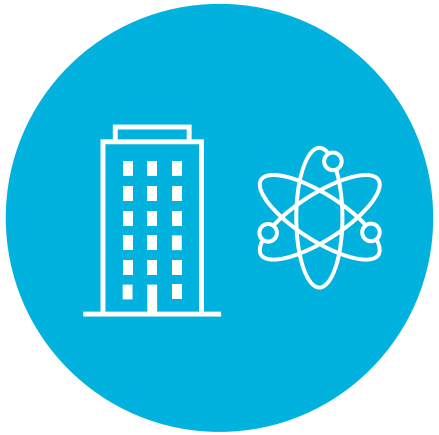
- include the creation of a **new data management system** which would collect real-world patient information to support optimal drug usage.
- serve as **reference centres** providing consultation and support to local healthcare practitioners allowing patients to receive optimal care close to home.



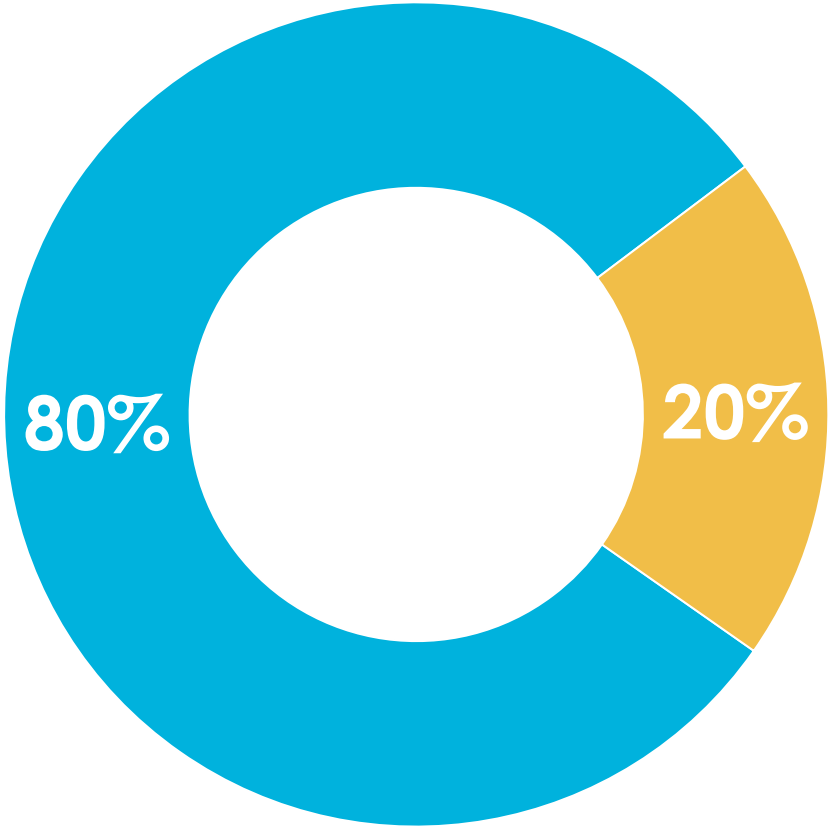
MODEL: NATIONAL LIST OF DRUGS

Funding would be allocated to provincial and federal public drug programs by subsidizing, or covering fully, a **defined national list of drugs** for those living with rare diseases. This approach means **all patients across Canada** would have the **same access to the drugs on the list regardless of where they live or whether they have a private drug plan**. However, this is based on the amount of budget allocated, the national list would include only a **limited number of approved drugs** and would **cover only selected diseases**.

Eight in 10 chose the rare disease centres over a national list of drugs as a preferred model for the national rare disease treatment strategy

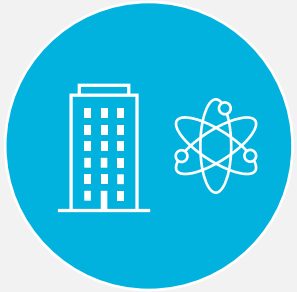


RARE DISEASE CENTRES is a better option because it provides **comprehensive support**, and the budget/ funding would be **allocated to both drugs and infrastructure** to promote timely diagnosis and optimal drug access for patients living with rare diseases **across Canada**.



NATIONAL LIST OF DRUGS is a better option because a **majority of the budget/funding will go to a defined list of Health Canada-approved drugs** for treating rare diseases, **regardless of where patients live in Canada**.

Q19. Keeping in mind the budget commitment for each model would be the same, which of the following best reflects your opinion?
Base: All respondents answering (n=370)



RARE DISEASE CENTRES

A centre with specialists of different backgrounds working on the same problem ***opens doors for collaboration and optimal results****.*

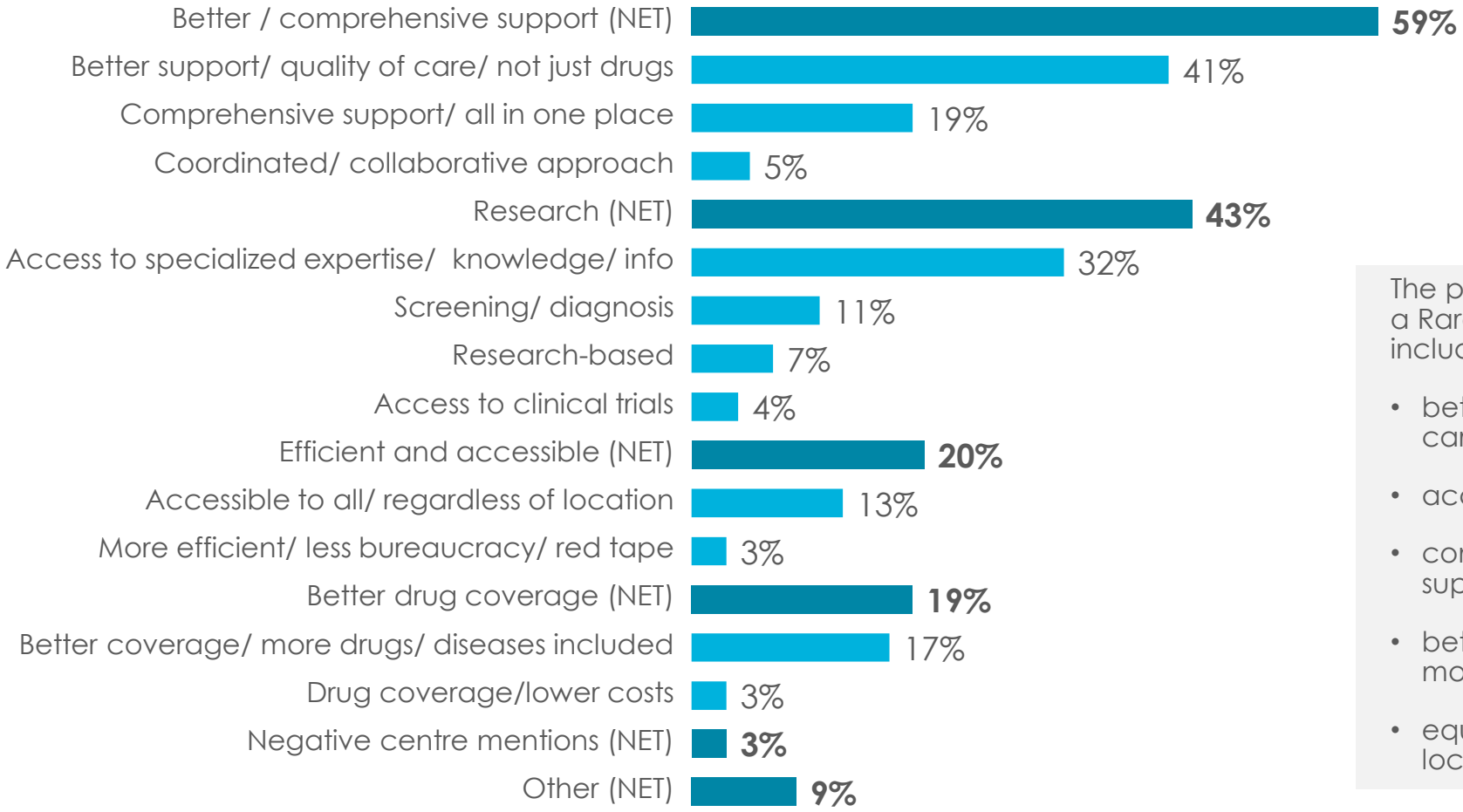
So we can ***deal with one place*** *and not run around to different doctors who aren't interested in helping*

There is a need for this kind of centre for ***access for ALL Canadians*** *regardless of the population of a province. Currently people in provinces with small populations have to travel to Ontario for treatment and diagnosis.* ***We can do better****.*

Rare diseases are treatable and a national strategy for a huge landmass with just over 40 million people makes sense.

We need health models that ***are uniform across Canada****, and because provincial politics are so subject to ideological influences and the imperatives of getting elected. I think most of all that funding and commitment has to be well supported and long-term. I also think* ***a national body has a better chance of attracting the brightest and most committed*** *to making lasting change.*

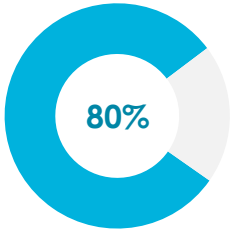
Favourability towards Rare Disease Centres Model



The primary reasons for favouring a Rare Disease Centres Model include:

- better support and quality of care
- access to specialized expertise
- comprehensive *one-stop-shop* support, and
- better coverage and drugs for more diseases
- equitable access regardless of location

Q20. Why did you choose Model: Rare Disease Centres? Please explain.
Base: All respondents selecting Rare Disease Centres model (n=253)



In Favour of Rare Disease Centres Model

BETTER / COMPREHENSIVE SUPPORT

*A centre with specialists of different backgrounds working on the same problem **opens doors for collaboration and optimal results**.*

*Treating a patient with a rare disease **goes beyond** access to rare and expensive drugs. Rare disease centres would **address a wide variety of gaps** that exist in the diagnosis and treatment of rare diseases.*

*My disease is not cured by medication and there is not enough specialists in the field. Having centers **makes more sense** in my case. I hate the idea of having a "pill to fix what ails you"...they usually cause other issues.*

BETTER DRUG COVERAGE

Drug shortages is already a crisis. Any potential shortage would be identified sooner.

*The coordinated centres for **better patient care** & information along with **clinical drug trial**s for rare diseases is the ideal. Of course, depending on where a patient might live, some might rely on **satellite centres** for some of their management.*

EFFICIENT AND ACCESSIBLE

*Our children need very little in the way of drugs. We need **early access to professionals** in the areas of genetic testing, speech therapy, dentistry, physiotherapy, behaviour management, respite services, sleep studies, epileptic care.*

*So we can **deal with one place** and not run around to different doctors who aren't interested in helping*

*The second model is **face to face care** (or telecare) which is much **more personal and patient specific**. EASY TO SAY NO IN THE FIRST MODEL, HARD TO SAY NO TO A REAL PERSON IN MODEL TWO.*

*There is a need for this kind of centre for **access for ALL Canadians** regardless of the population of a province. Currently people in provinces with small populations have to travel to Ontario for treatment and diagnosis. **We can do better**. Rare diseases are treatable and a national strategy for a huge landmass with just over 40 million people makes sense.*

*Rare disease diagnosis and management requires **optimal** and **up to date expertise** in both clinical care and research. This model also permits **real world data tracking** of patient experience using new therapies that are approved but perhaps not formally offered based on rigid criteria, thereby expanding data available for new drugs following clinical trials.*

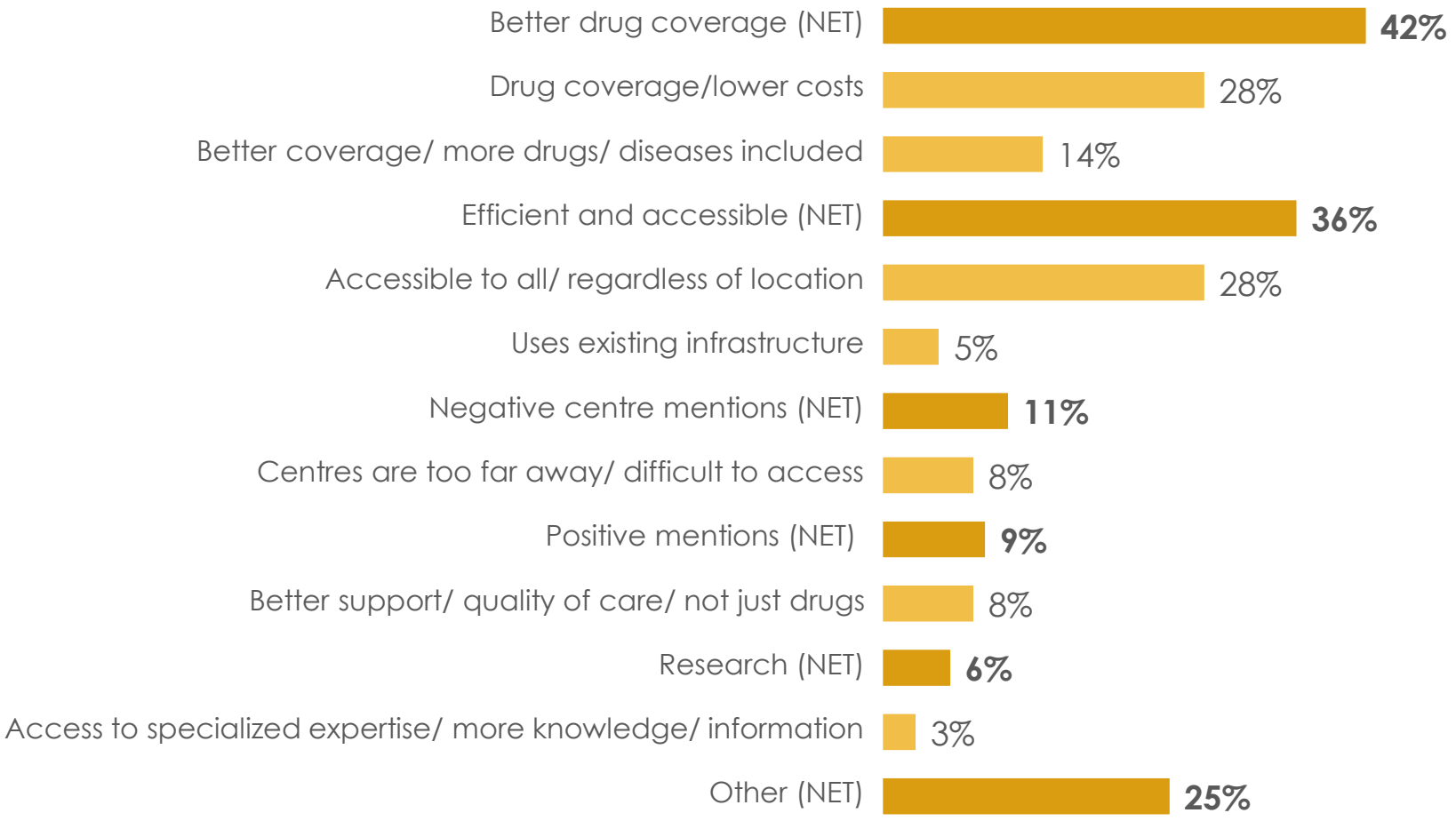
RESEARCH

*I would like to see more support for **research** and **clinical trials** in Canada.*

*Don't like a defined list (**risk for too many exclusions**). Like best practice model. Like experts available and research investments.*

Q20. Why did you choose Model: Rare Disease Centres? Please explain.
Base: All respondents selecting Rare Disease Centres model (n=253)

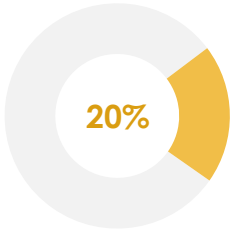
Favourability towards National List of Drugs Model



The primary reasons for favouring a National List of Drugs model include:

- Lower costs on drug coverage
- equitable access to drugs regardless of location
- better coverage and drugs for more diseases

Q20. Why did you choose Model: National List of Drugs? Please explain.
Base: All respondents selecting National List of Drugs model (n=64)



In Favour of National List of Drugs Model

BETTER DRUG COVERAGE

*Because it would be **accessible by everyone** – no matter where they lived. Also, with the other – there would be a lot of funding money going into infrastructure – which would be in big cities – not in the remote areas.*

*We have infrastructure that we can use across Canada. I support research, yes, but **not everyone in Canada will be able to access these sites** and there will be inequities in access to the necessary drugs.*

*The centres of excellence are limited by the **referral process** to them. Their existence won't immediately change physician referral processes or practice. The **geographic distances in Canada limit the accessibility** to these centres.*

*Funding for existing drugs will **have immediate implications** for currently affected patients and should be prioritized.*

EFFICIENT AND ACCESSIBLE

*This model is not based on where someone lives and the **drug costs are currently my priority** versus the infrastructure.*

*Because **cost is the biggest factor for me**, I spend close to 500 a year in meds and it's a huge cost for someone making minimum wage*

*The **comprehensive centres would not be accessible to all Canadians**. A person with the rare disease is **not mobile or able to travel**. Remote access would also be very hard as my mother lost her ability to speak early in her disease.*

POSITIVE MENTIONS

*Drugs especially for rare disease continue to get more and more expensive. In my experience, the acute diagnosis and treatment I received at a traditional hospital was quite good. Assuming that everyone can access a hospital in a major center then specialists in the hospitals should be able to diagnose most rare diseases. This would leave the bulk of funding the actual medicines**. I worry if funding is split between medicine and infrastructure then some drugs won't be covered.** Ultimately, I feel its the drugs that people need access to and therefore the **plan which provides the greatest number of drugs to funded is best.***

CHALLENGES WITH RARE DISEASE CENTRES

*Much more expedient in having it up and running as opposed to clinics which **will be difficult to staff, manage and accommodate** for the numbers of clients.*

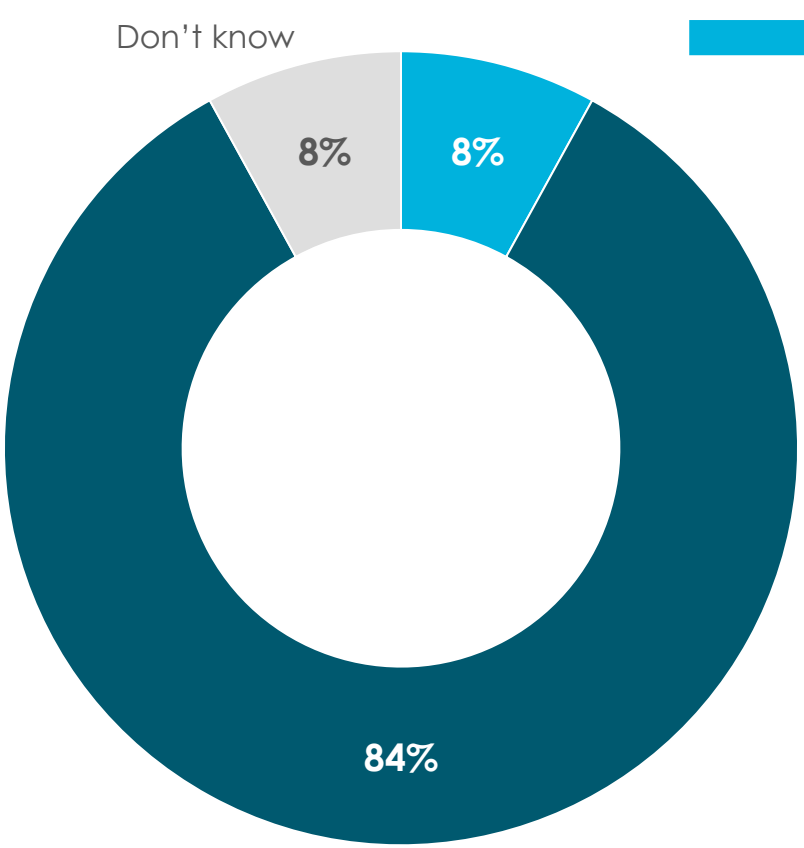
*Because **I doubt a centre would be located near me** anyway*

***travelling** and access to locations/centers **will be challenging** for the patient*

*It will be **impossible for different centres to offer the same level of care**, I think it will devolve into some high quality, some medium, some low*

Q20. Why did you choose Model: National List of Drugs? Please explain.
Base: All respondents selecting National List of Drugs model (n=64)

Majority lean towards federal oversight as the government model that would improve access to treatment and care for those living with rare diseases; highest in Alberta.



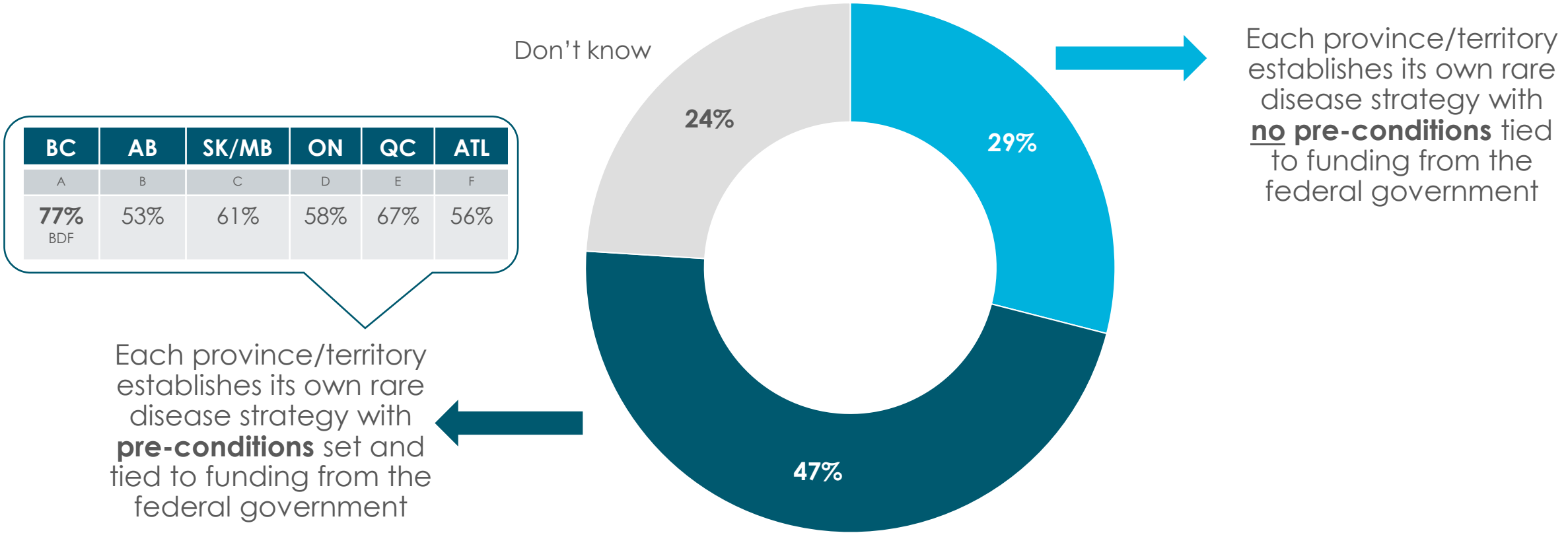
Each province/ territory establishes and runs its own rare disease strategy

BC	AB	SK/MB	ON	QC	ATL
A	B	C	D	E	F
91%	**100%** ADE	97%	90%	84%	95%

Federal government would provide funding, support and standards for a coordinated inter-provincial rare disease strategy

Q22. Which of the following government models do you think is the best way to improve access to treatment and care for those living with rare diseases?
Base: All respondents answering (n=516)

Almost half of patients prefer provinces to have pre-set conditions tied to federal funding; highest in BC.



BC	AB	SK/MB	ON	QC	ATL
A	B	C	D	E	F
77% BDF	53%	61%	58%	67%	56%

Q23. If each province/territory is provided the funding to spend, which model would you support?
Base: All respondents answering (n=519)

PROFILE OF SURVEY RESPONDENTS AND INTERVIEWEES

Phase 1: Profile of Patients with Rare Diseases

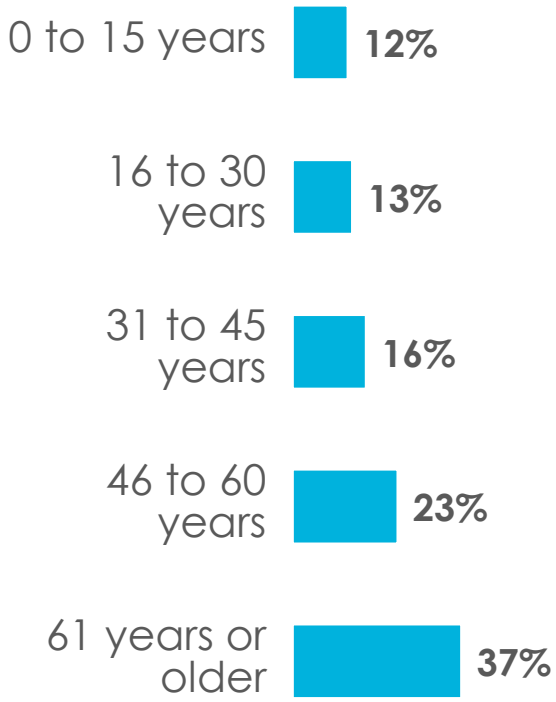
WHO IS ANSWERING SURVEY

72%
PATIENT with a rare disease

28%
CAREGIVER

Q1. Who is answering this questionnaire?
Base: All respondents answering (n=528)

AGE GROUPS OF PATIENTS



Q2. In what year was the person with the rare disease born?
Base: All respondents answering (n=524)

GENDER OF RARE DISEASE PATIENT

69%
Woman

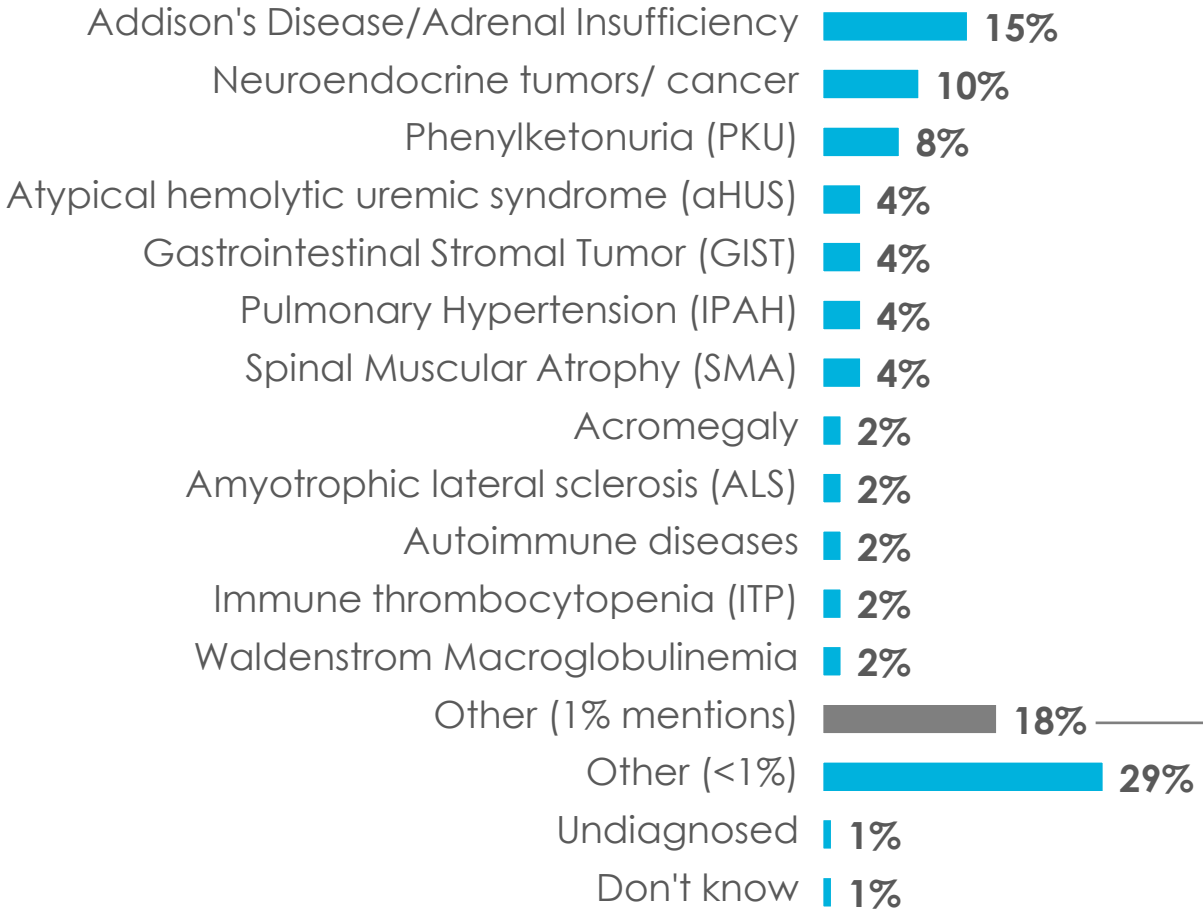
29%
Man

2%
Transgender/ non-binary/ non-conforming/ another

1%
Prefer not to answer

Q3. How does the person with a rare disease identify?
Base: All respondents answering (n=528)

Name of Rare Disease



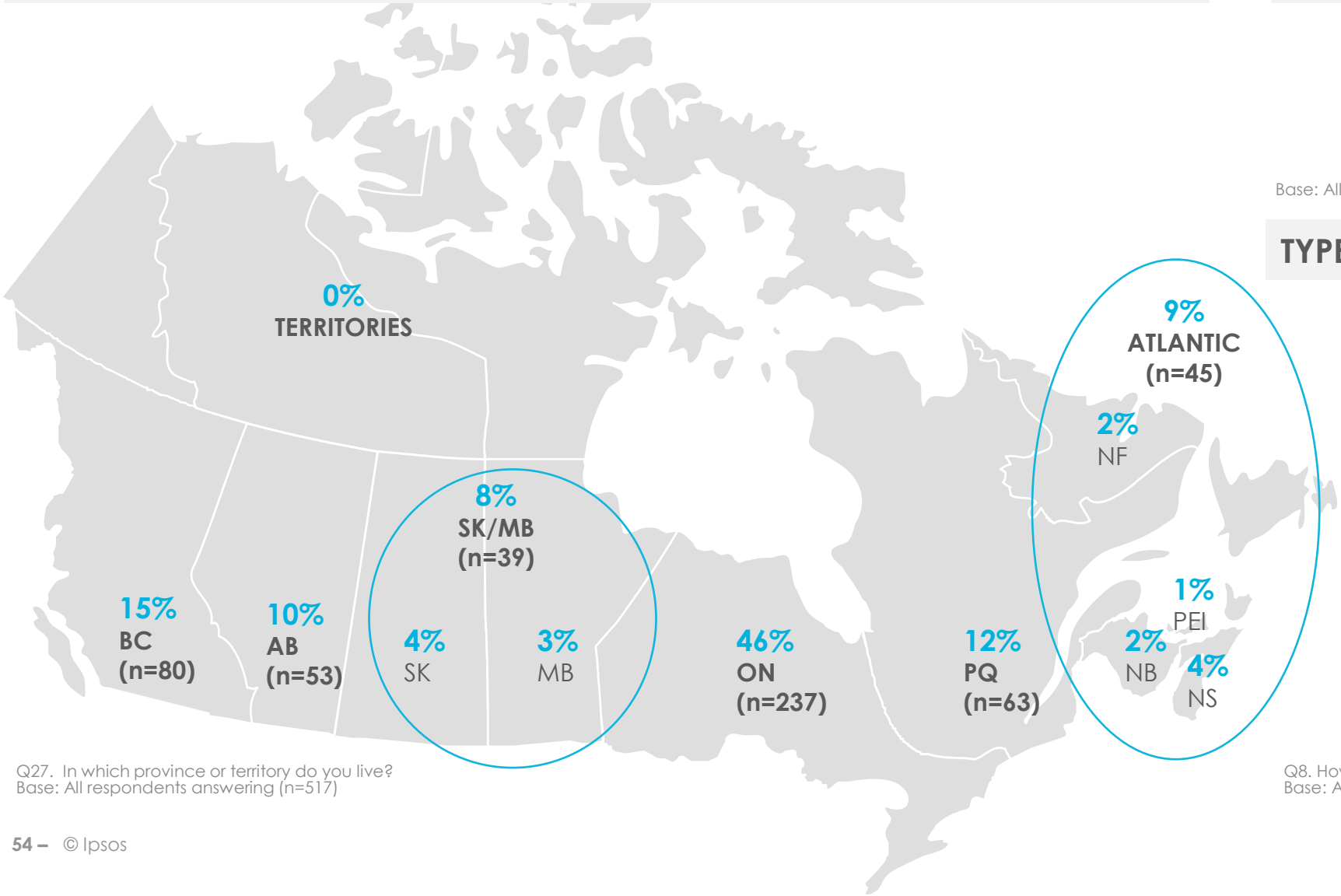
Mentions of 1%

- CDKL5 disorder (CDD)
- Congenital Adrenal Hyperplasia
- Cystic Fibrosis (CF)
- Diffuse idiopathic pulmonary neuroendocrine cell hyperplasia (DIPNECH)
- Dravet syndrome
- Ehlers-Danlos syndrome
- Fabry Disease
- Hereditary Angioedema
- Hypogammaglobulinemia
- Liposarcoma
- Loeys-Dietz syndrome
- Myasthenia gravis
- Rett Syndrome
- Schmidt syndrome
- Von Hippel-Lindau disease (VHL)
- X-Linked Hypophosphatemia (XLH)

Q4. What is the name of the rare disease?
Base: All respondents answering (n=525)

Geographic distribution

PROVINCE/TERRITORY RESIDE IN



Q27. In which province or territory do you live?
Base: All respondents answering (n=517)

LANGUAGE

90% English

10% French

Base: All respondents answering (n=528)

TYPE OF AREA RESIDE IN

47% Urban

34% Suburban

20% Rural

Q8. How would you classify the area in which you currently live?
Base: All respondents answering (n=512)

Phase 1: Profile of Caregivers

- Of the 28% respondents who are caregivers of someone with a rare disease, majority are women and older than 31 years old.

WHO IS ANSWERING SURVEY

72%
PATIENT with a rare disease

28%
CAREGIVER

Q1. Who is answering this questionnaire?
Base: All respondents answering (n=528)

GENDER

85%
Woman

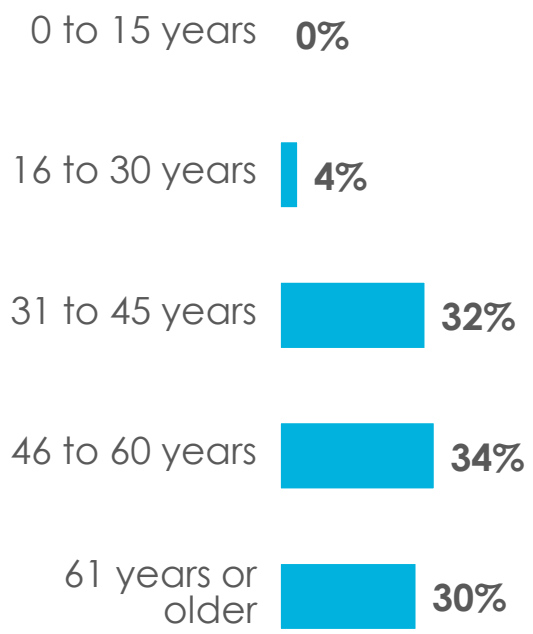
14%
Man

1%
Transgender/non-binary/ non-conforming/another

1%
Prefer not to answer

Q25. Do you identify as …?
Base: All respondents answering (n=144)

AGE



Q26. In what year were you born?
Base: All respondents answering (n=140)

Phase 2: Profile of Participants

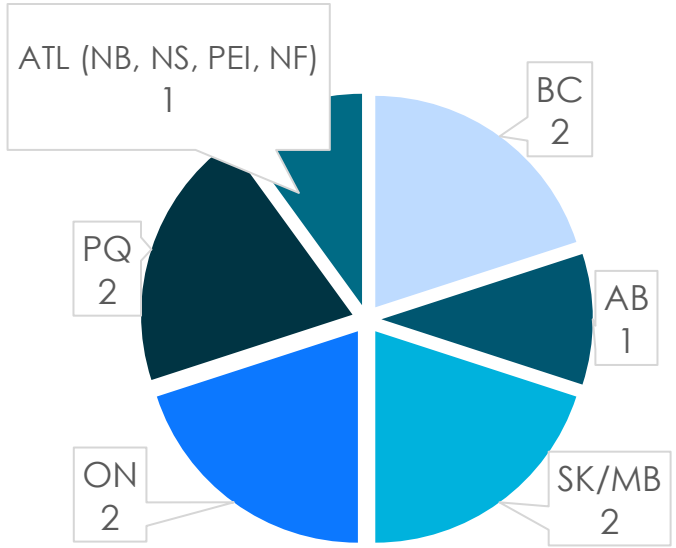
WHO WAS INTERVIEWED

7
PATIENTS with a rare disease

3
CAREGIVERS

- Parent
- Spouse

REGION OF PATIENT AND FAMILY



4 Urban

4 Suburban

2 Rural

GENDER OF RARE DISEASE PATIENT

6
Woman

4
Man

RARE DISEASE GENETICS

4
Genetic/hereditary

3
Not genetic/inherited

3
Unsure

Base: All respondents (n=10)

Contact

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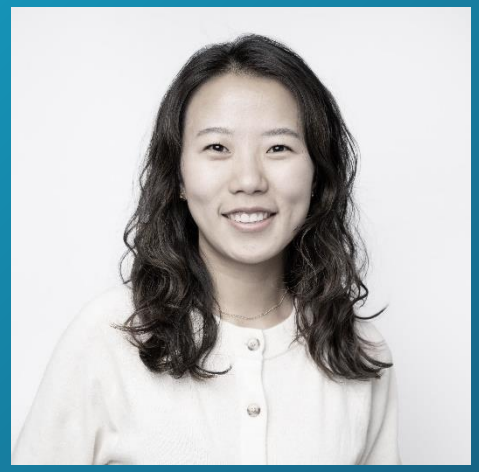

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