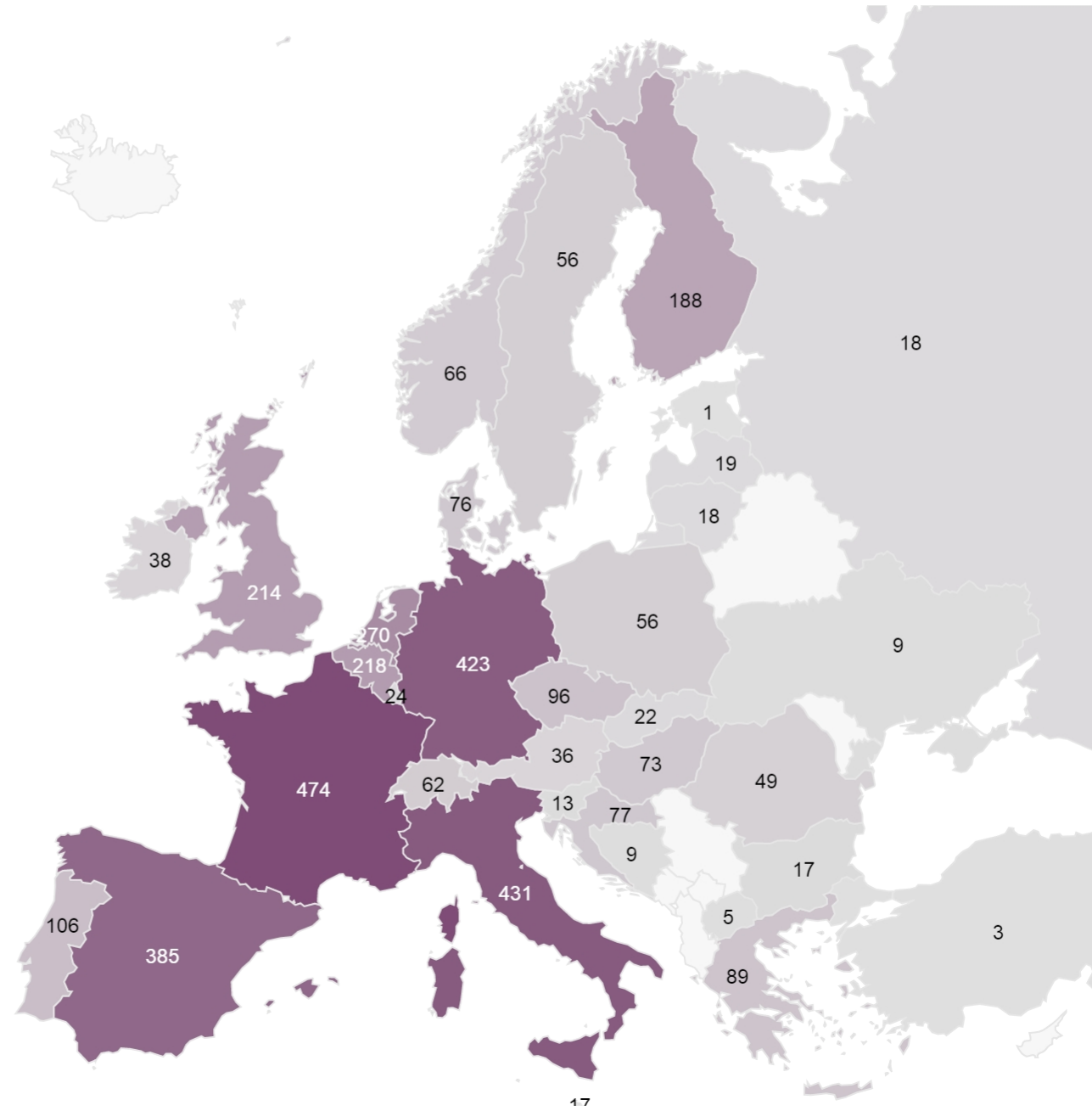
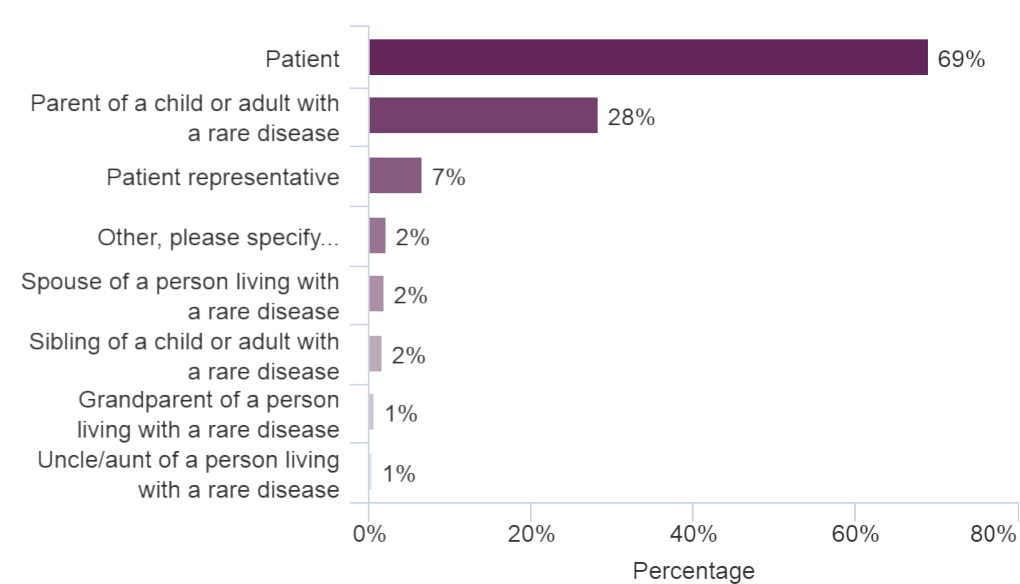


### In which country do you live?



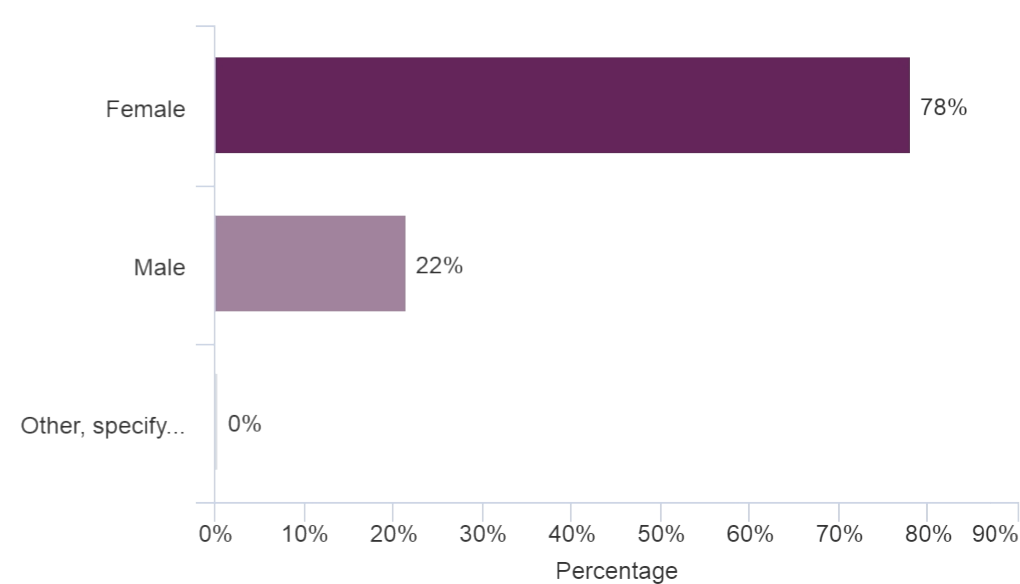
How would you like to answer this questionnaire? In your capacity as a ... Several answers possible

	N
Patient	2,536
Parent of a child or adult with a rare ...	1,038
Patient representative	243
Other, please specify...	81
Spouse of a person living with a rare...	68
Sibling of a child or adult with a rare ...	64
Grandparent of a person living with ...	27
Uncle/aunt of a person living with a r...	21
TOTAL	3,662



Are you :

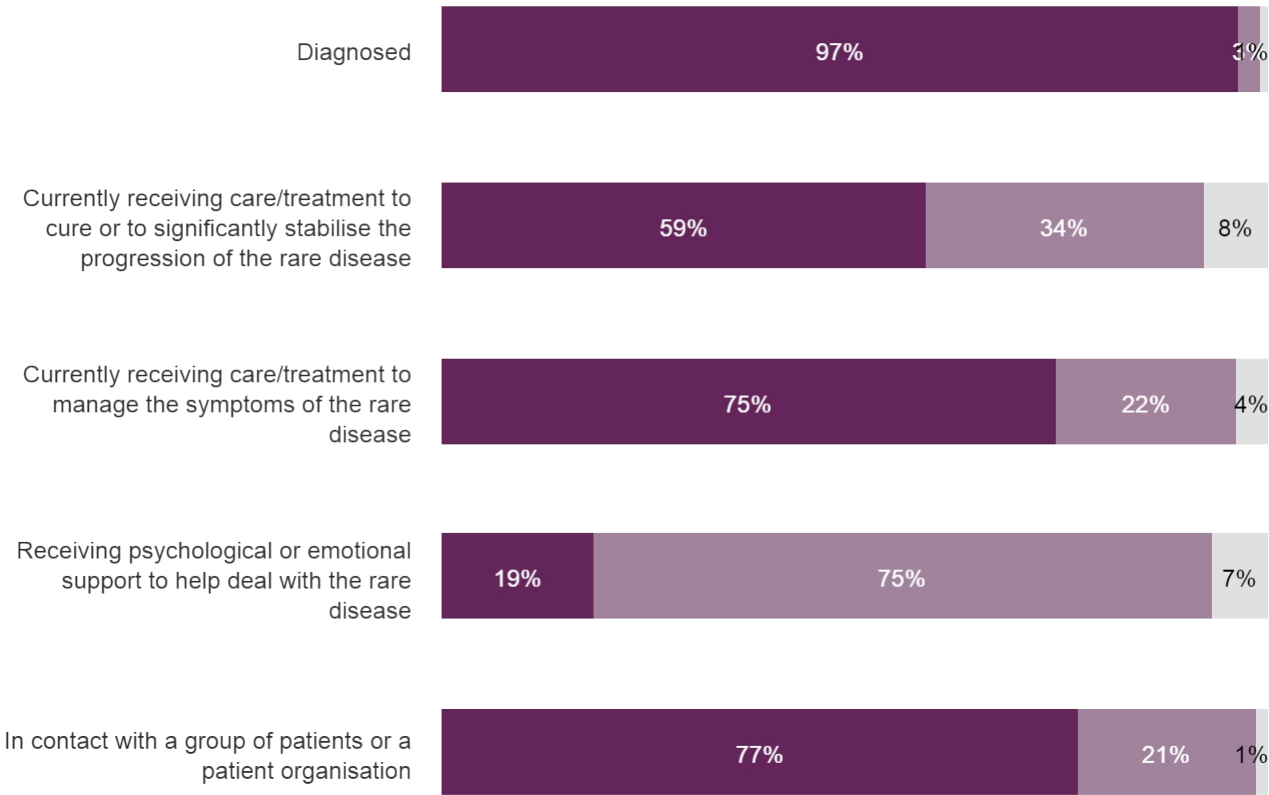
	N
Female	2,820
Male	779
Other, specify...	14
TOTAL	3,613



Are you or the person you care for:

	YES	NO	NOT APPLICABLE	TOTAL
Diagnosed	3,539	96	27	3,662
Currently receiving care/treatment to cure or to significantly stabilise the progression of the rare disease	2,154	1,231	277	3,662
Currently receiving care/treatment to manage the symptoms of the rare disease	2,729	798	135	3,662
Receiving psychological or emotional support to help deal with the rare disease	686	2,737	239	3,662
In contact with a group of patients or a patient organisation	2,829	786	47	3,662
TOTAL	11,937	5,648	725	18,310

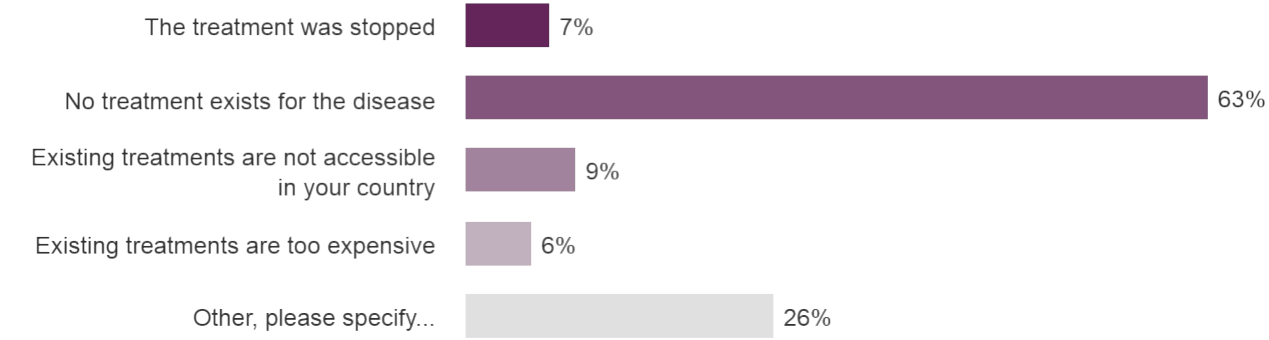
Are you or the person you care for:



You or the person you care for are not receiving treatment for the rare disease because:

	N
The treatment was stopped	119
No treatment exists for the disease	1,023
Existing treatments are not accessible in your country	153
Existing treatments are too expensive	93
Other, please specify...	176

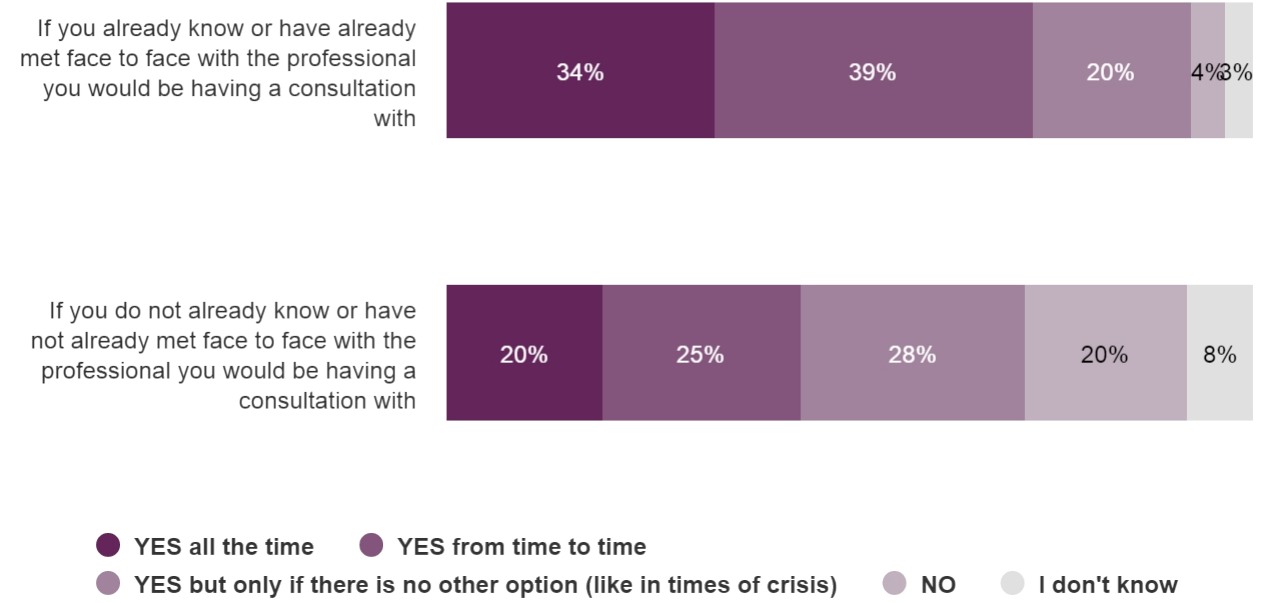
You or the person you care for are not receiving treatment for the rare disease because:



Within the next 10 years, would you be willing to attend remote consultations (via phone, video, email, text, app):

	YES ALL THE TIME	YES FROM TIME TO TIME	YES BUT ONLY IF THERE IS NO OTHER OPTION (LIKE IN TIMES OF CRISIS)	NO	I DON'T KNOW	TOTAL
If you already know or have already met face to face with the professional you would be having a consultation with	1,227	1,444	715	161	115	3,662
If you do not already know or have not already met face to face with the professional you would be having a consultation with	715	901	1,016	742	288	3,662
TOTAL	1,942	2,345	1,731	903	403	7,324

Within the next 10 years, would you be willing to attend remote consultations (via phone, video, email, text, app):

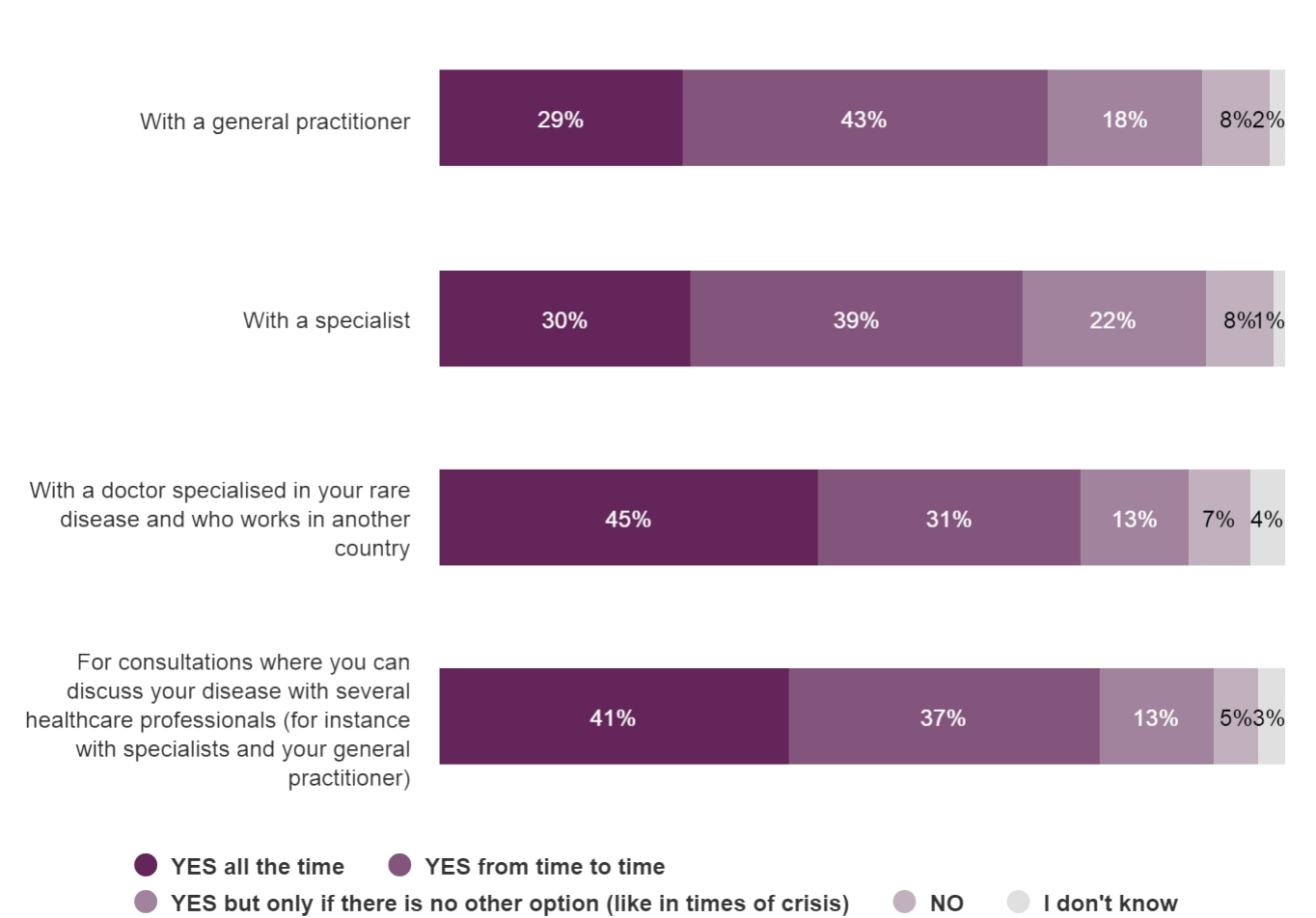




Within the next 10 years, would you be willing to attend remote consultations (via phone, video, email, text, app):

	YES ALL THE TIME	YES FROM TIME TO TIME	YES BUT ONLY IF THERE IS NO OTHER OPTION (LIKE IN TIMES OF CRISIS)	NO	I DON'T KNOW	TOTAL
With a general practitioner	1,060	1,581	674	289	58	3,662
With a specialist	1,096	1,434	800	289	43	3,662
With a doctor specialised in your rare disease and who works in another country	1,650	1,130	471	270	141	3,662
For consultations where you can discuss your disease with several healthcare professionals (for instance with specialists and your general practitioner)	1,518	1,353	490	189	112	3,662
TOTAL	5,324	5,498	2,435	1,037	354	14,648

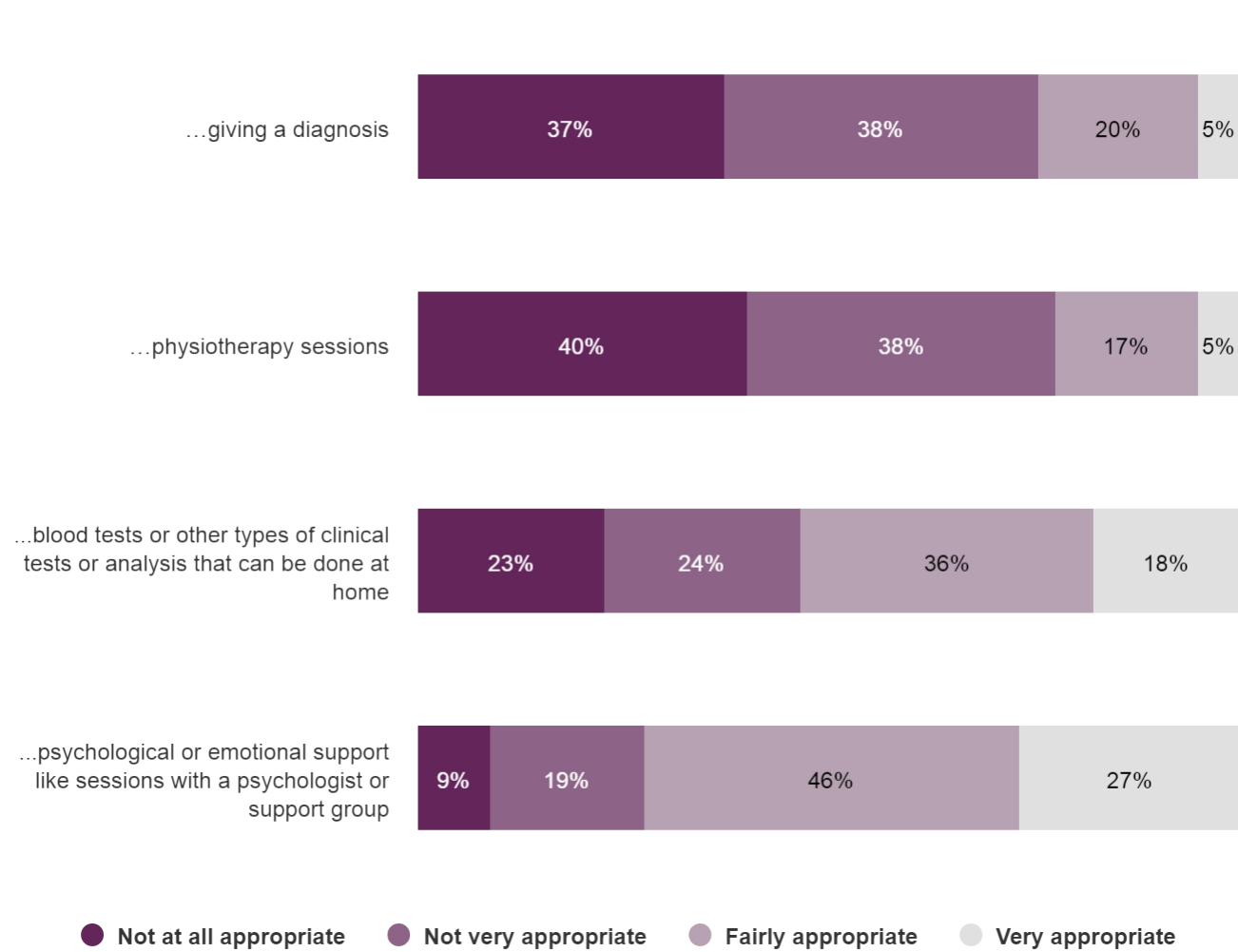
Within the next 10 years, would you be willing to attend remote consultations (via phone, video, email, text, app):



Do you think that remote virtual consultations are appropriate for...

	NOT AT ALL APPROPR...	NOT VERY APPROPR...	FAIRLY APPROPR...	VERY APPROPR...	TOTAL
...giving a diagnosis	1,340	1,362	701	172	3,575
...physiotherapy sessions	1,396	1,304	607	165	3,472
...blood tests or other types of clinical tests or analysis that can be done at home	799	829	1,242	613	3,483
...psychological or emotional support like sessions with a psychologist or support group	314	654	1,582	922	3,472
TOTAL	3,849	4,149	4,132	1,872	14,002

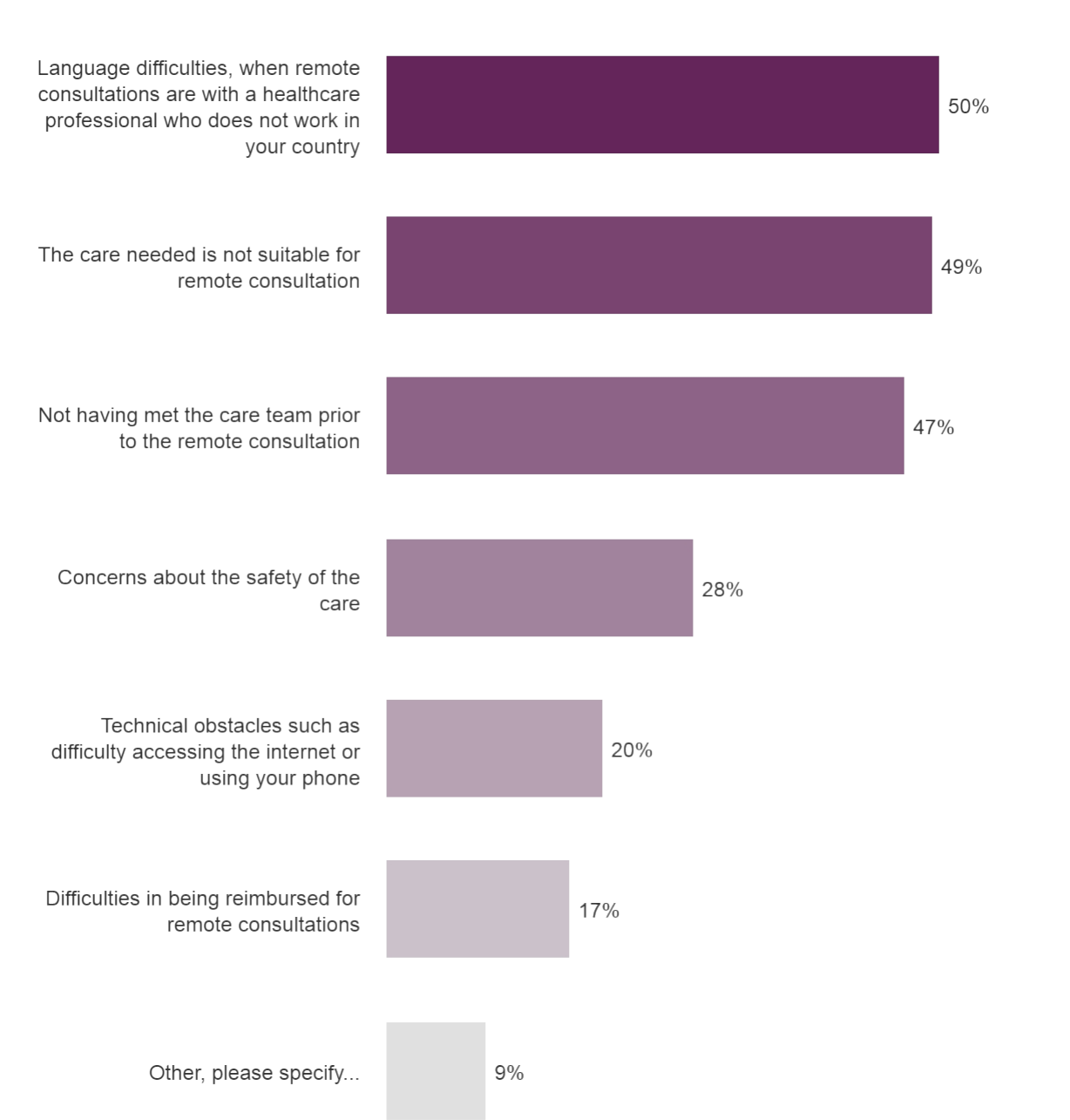
Do you think that remote virtual consultations are appropriate for...



What would be the 3 main obstacles for you to use remote consultations within the next 10 years? 3 answers maximum

	N
Language difficulties, when remote consultations are with a healthcare professional who does not work in your country	1,824
The care needed is not suitable for remote consultation	1,803
Not having met the care team prior to the remote consultation	1,709
Concerns about the safety of the care	1,016
Technical obstacles such as difficulty accessing the internet or using your phone	717
Difficulties in being reimbursed for remote consultations	606
Other, please specify...	331
TOTAL	3,662

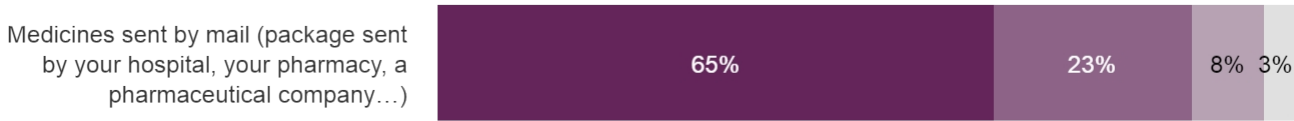
What would be the 3 main obstacles for you to use remote consultations within the next 10 years? 3 answers maximum



Within the next 10 years, would you be willing to use the following services:

	YES ALL THE TIME	YES BUT ONLY IF THERE IS NO OTHER OPTION (LIKE IN TIMES OF CRISIS)	NO	I DON'T KNOW	TOTAL
Prescriptions sent by email (electronic message)	3,075	449	78	60	3,662
Prescriptions sent by mail (posted letter)	1,584	879	1,059	140	3,662
Medicines sent by mail (package sent by your hospital, your pharmacy, a pharmaceutical company...)	2,391	847	301	123	3,662
TOTAL	7,050	2,175	1,438	323	10,986

Within the next 10 years, would you be willing to use the following services:

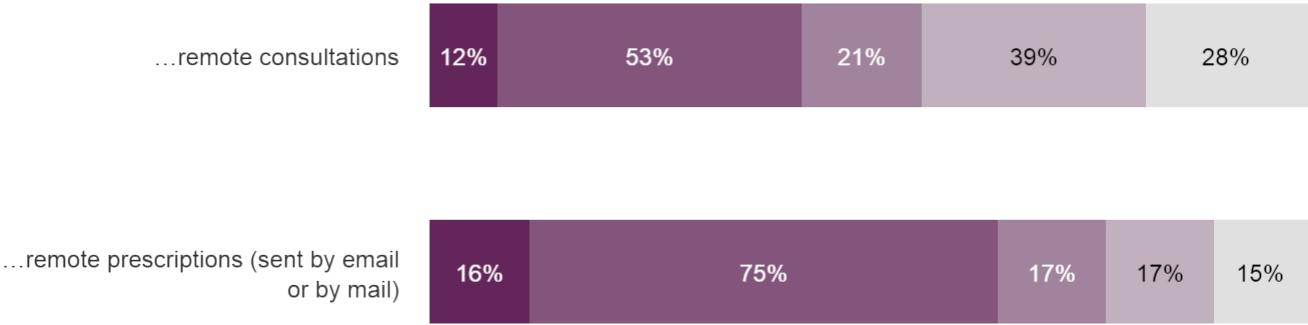


● YES all the time    ● YES but only if there is no other option (like in times of crisis)    ● NO  
● I don't know

Within the next 10 years, what would be the two main reasons for you to use...

	SAVING MONEY	SAVING TIME	SAFETY	ACCES... HIGHER QUALITY CARE	HAVING NO OTHER OPTION	TOTAL
...remote consultations	440	1,938	767	1,415	1,019	5,579
...remote prescriptions (sent by email or by mail)	600	2,738	638	630	539	5,145
TOTAL	1,040	4,676	1,405	2,045	1,558	10,724

Within the next 10 years, what would be the two main reasons for you to use...



- Saving money
- Saving time
- Safety
- Accessing higher quality care
- Having no other option

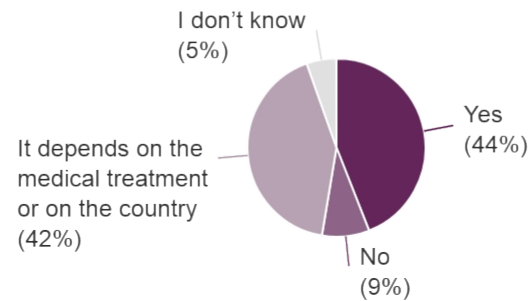
Within the next 10 years and OUTSIDE TIMES OF CRISES, would you be willing to travel to another country in the European Union to receive medical treatment for your rare disease?

	N
Yes	1,441
No	281
It depends on the medical treatment or on the country	1,373
I don't know	177
TOTAL	3,272

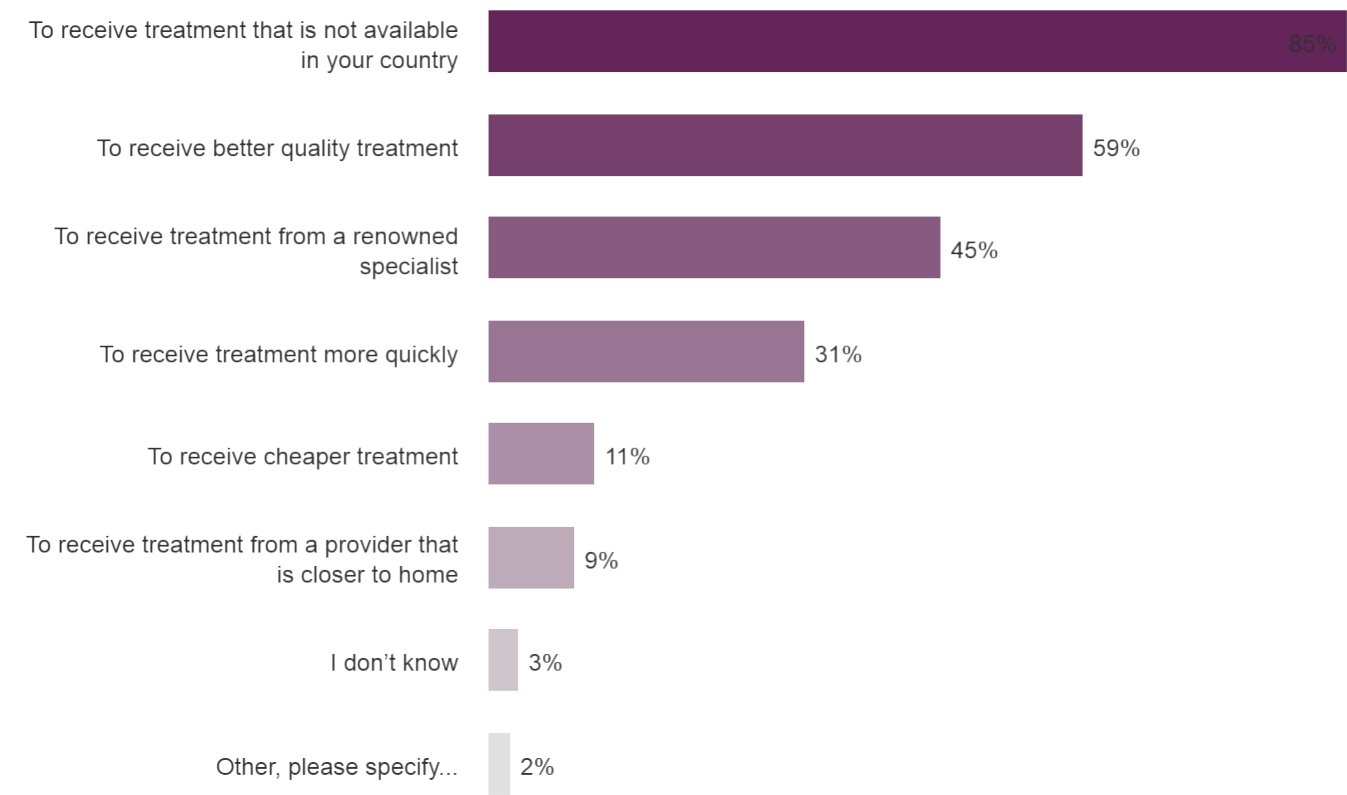
For which of the following reasons would you be willing to travel to another country in the European Union to receive medical treatment for your rare disease? Several answers possible

	N
To receive treatment that is not available in your country	2,541
To receive better quality treatment	1,762
To receive treatment from a renowned specialist	1,339
To receive treatment more quickly	939
To receive cheaper treatment	320
To receive treatment from a provider that is closer to home	261
I don't know	94
Other, please specify...	68
TOTAL	2,991

Within the next 10 years and OUTSIDE TIMES OF CRISES, would you be willing to travel to another country in the European Union to receive medical treatment for your rare disease?



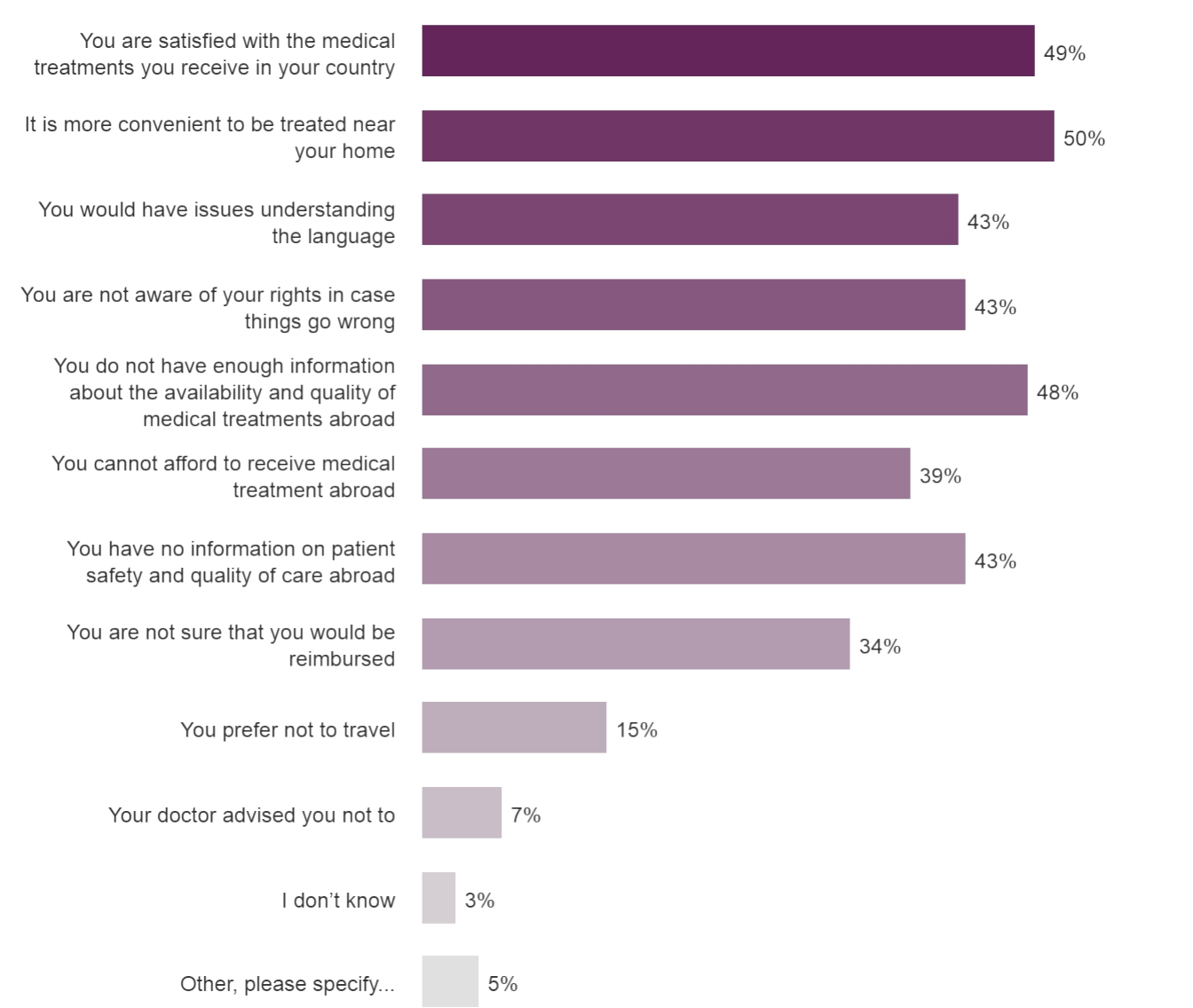
For which of the following reasons would you be willing to travel to another country in the European Union to receive medical treatment for your rare disease? Several answers possible



For which of the following reasons would you not be willing to go to another country in the European Union to receive medical treatment for your rare disease(s)? Several answers possible

	N
You are satisfied with the medical treatments you receive in your country	895
It is more convenient to be treated near your home	924
You would have issues understanding the language	783
You are not aware of your rights in case things go wrong	794
You do not have enough information about the availability and quality of medical treatments abroad	885
You cannot afford to receive medical treatment abroad	713
You have no information on patient safety and quality of care abroad	794
You are not sure that you would be reimbursed	626
You prefer not to travel	272
Your doctor advised you not to	120
I don't know	52
Other, please specify...	85
TOTAL	1,831

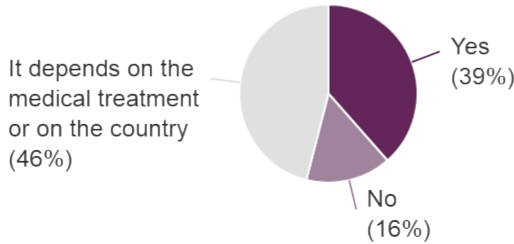
For which of the following reasons would you not be willing to go to another country in the European Union to receive medical treatment for your rare disease(s)? Several answers possible



Within the next 10 years and OUTSIDE TIMES OF CRISES, would you be willing to travel to another country to receive medical treatment for your rare disease?

	N
Yes	144
No	58
It depends on the medical treatment or on the country	172
I don't know	16
TOTAL	390

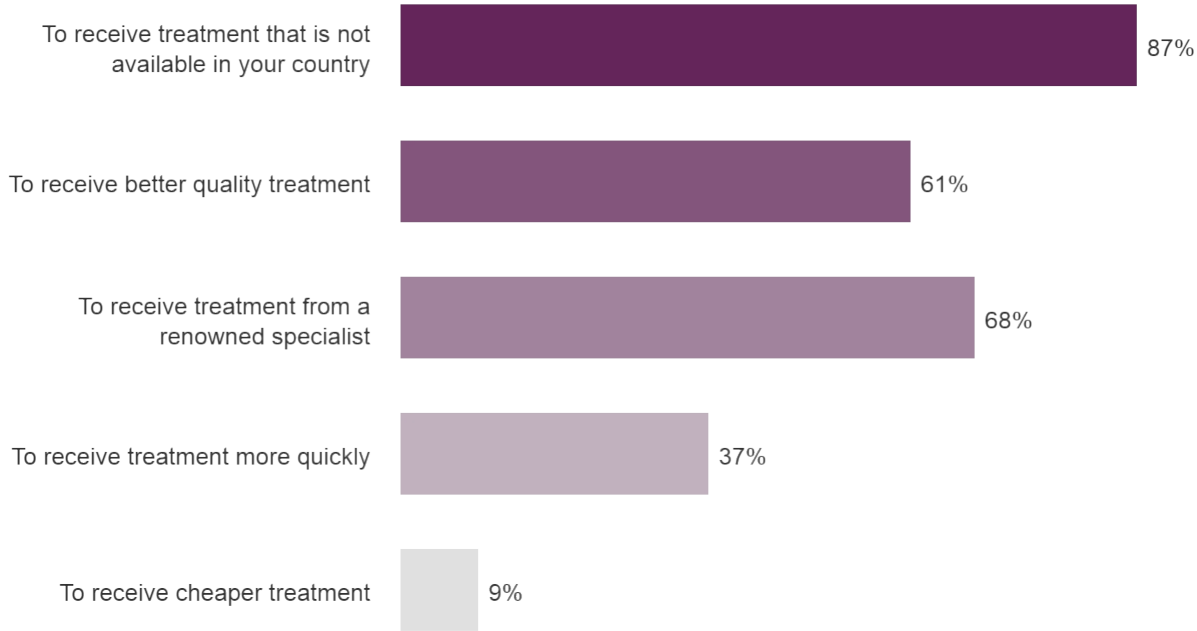
Within the next 10 years and OUTSIDE TIMES OF CRISES, would you be willing to travel to another country to receive medical treatment for your rare disease?



For which of the following reasons would you be willing to travel to another country to receive medical treatment for your rare disease? Several answers possible

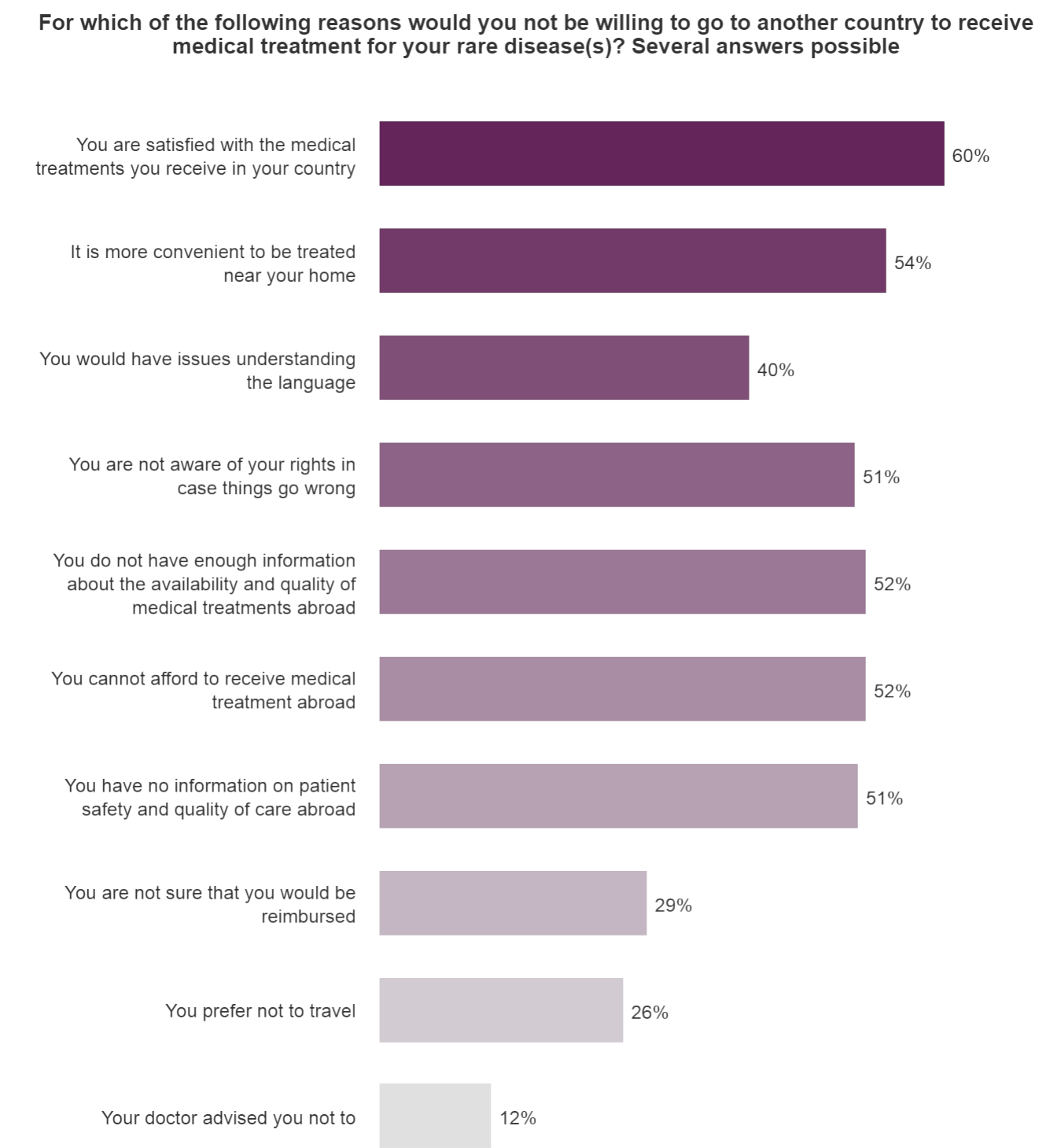
	N
To receive treatment that is not available in your country	276
To receive better quality treatment	192
To receive treatment from a renowned specialist	216
To receive treatment more quickly	116
To receive cheaper treatment	30
To receive treatment from a provider that is closer to home	14
I don't know	13
Other, please specify...	10
TOTAL	332

For which of the following reasons would you be willing to travel to another country to receive medical treatment for your rare disease? Several answers possible





For which of the following reasons would you not be willing to go to another country to receive medical treatment for your rare disease(s)? Several answers possible	
	N
You are satisfied with the medical treatments you receive in your country	145
It is more convenient to be treated near your home	130
You would have issues understanding the language	95
You are not aware of your rights in case things go wrong	122
You do not have enough information about the availability and quality of medical treatments abroad	125
You cannot afford to receive medical treatment abroad	125
You have no information on patient safety and quality of care abroad	123
You are not sure that you would be reimbursed	69
You prefer not to travel	63
Your doctor advised you not to	29
I don't know	4
Other, please specify...	16
TOTAL	246



Here are some statements related to health care received in another country of the European Union. For each of the following, could you please say whether you think it is true or false?

	TRUE	FALSE	I DON'T KNOW	TOTAL
You have the right to receive planned medical treatment in another country in the European Union and to be reimbursed for that treatment by national health authority or healthcare insurer.	1,433	496	1,343	3,272
You cannot get a prescription from your doctor to use in another country in the European Union.	905	845	1,522	3,272
You have the right to receive a copy of your medical record from your doctor when you seek to receive healthcare in another country in the European Union.	2,471	57	744	3,272
Your doctor may be able to get help in treating you locally by contacting a network of doctors specialised in specific rare disease(s) called a European Reference Network.	1,841	105	1,326	3,272
TOTAL	6,650	1,503	4,935	13,088

Here are some statements related to health care received in another country of the European Union. For each of the following, could you please say whether you think it is true or false?



Do you think it is possible and realistic that within 10 years, you or the person you care for...

	THIS IS ALREADY THE CASE	YES AND THIS IS NOT YET THE CASE	NO	I DON'T KNOW	TOTAL
...could be cured from the rare disease: total remission, no symptoms.	79	565	2,419	599	3,662
...could have their rare disease stabilised: the rare disease would still be present but symptoms would not be progressing.	672	1,596	728	666	3,662
...could manage the symptoms of the rare disease even if they are still progressing.	925	1,363	504	870	3,662
...could be supported to manage the psychological or emotional aspects of the rare disease.	925	1,712	309	716	3,662
TOTAL	2,601	5,236	3,960	2,851	14,648

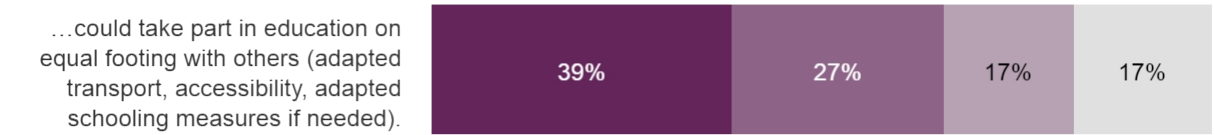
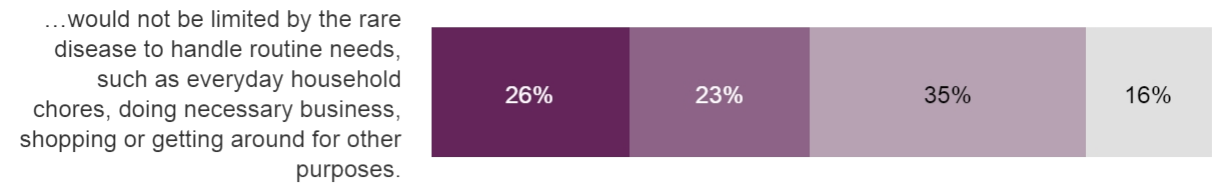
Do you think it is possible and realistic that within 10 years, you or the person you care for...



● This is already the case   ● YES and this is not yet the case   ● NO   ● I don't know

Do you think it is possible and realistic that within 10 years, you or the person you care for...	THIS IS ALREADY THE CASE	YES AND THIS IS NOT YET THE CASE	NO	I DON'T KNOW	TOTAL
...would not be limited by the rare disease to handle routine needs, such as everyday household chores, doing necessary business, shopping or getting around for other purposes.	941	843	1,295	583	3,662
...could take part in education on equal footing with others (adapted transport, accessibility, adapted schooling measures if needed).	1,414	1,000	611	637	3,662
...could access adapted and accessible employment as well as flexible work arrangements (adjustment of working hours, working remotely).	886	1,320	795	661	3,662
...would not be discriminated against due to their rare disease or due to their disabilities, in the various aspects of their daily life.	844	1,170	963	685	3,662
TOTAL	4,085	4,333	3,664	2,566	14,648

Do you think it is possible and realistic that within 10 years, you or the person you care for...

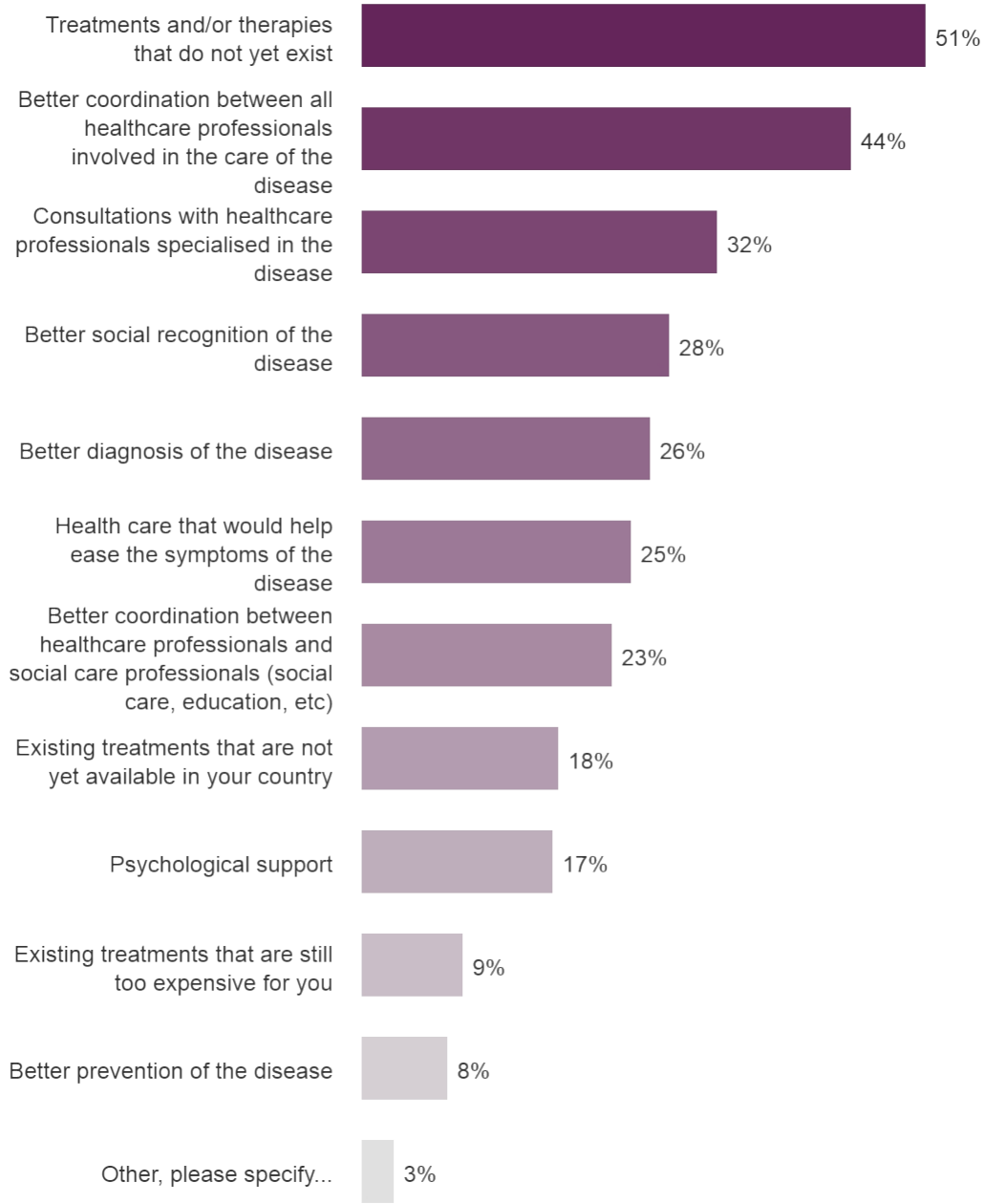


● This is already the case    ● YES and this is not yet the case    ● NO    ● I don't know

Within the next 10 years, the top 3 priorities to improve care for your rare disease would be to access to

	N
Treatments and/or therapies that do not yet exist	1,871
Better coordination between all healthcare professionals involved in the care of the disease	1,625
Consultations with healthcare professionals specialised in the disease	1,178
Better social recognition of the disease	1,024
Better diagnosis of the disease	962
Health care that would help ease the symptoms of the disease	899
Better coordination between healthcare professionals and social care professionals (social care, education, etc)	833
Existing treatments that are not yet available in your country	654
Psychological support	637
Existing treatments that are still too expensive for you	338
Better prevention of the disease	292
Other, please specify...	111
TOTAL	3,662

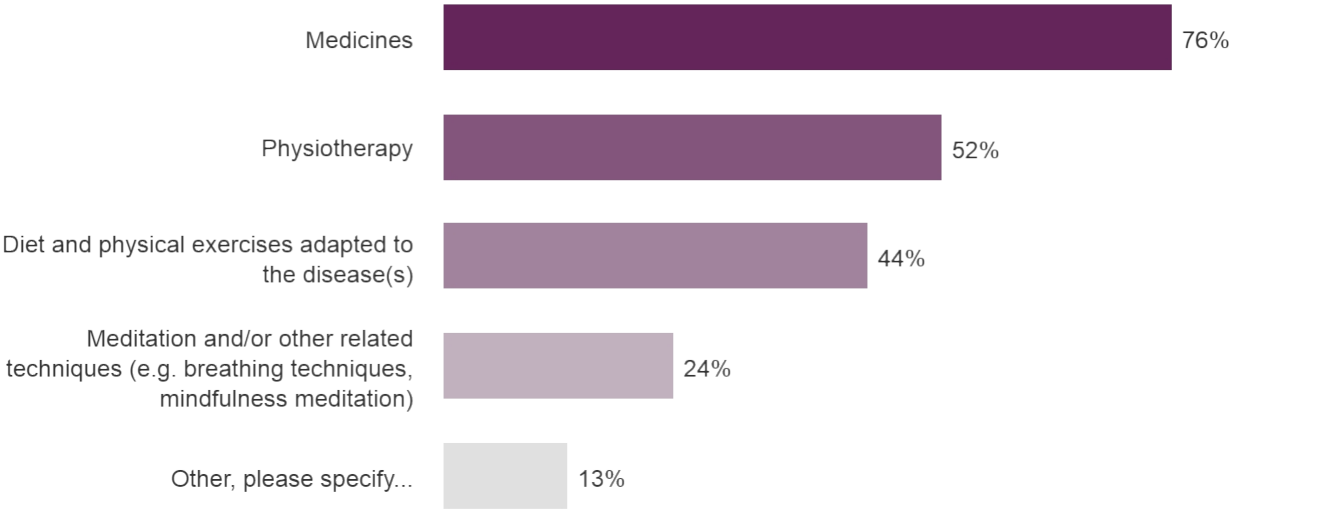
Within the next 10 years, the top 3 priorities to improve care for your rare disease would be to access to



Within the next 10 years, in order to help ease the symptoms of your rare disease, you would need access to:

	N
Medicines	685
Physiotherapy	469
Diet and physical exercises adapted to the disease(s)	400
Meditation and/or other related techniques (e.g. breathing techniques, mindfulness meditation)	218
Other, please specify...	119
TOTAL	899

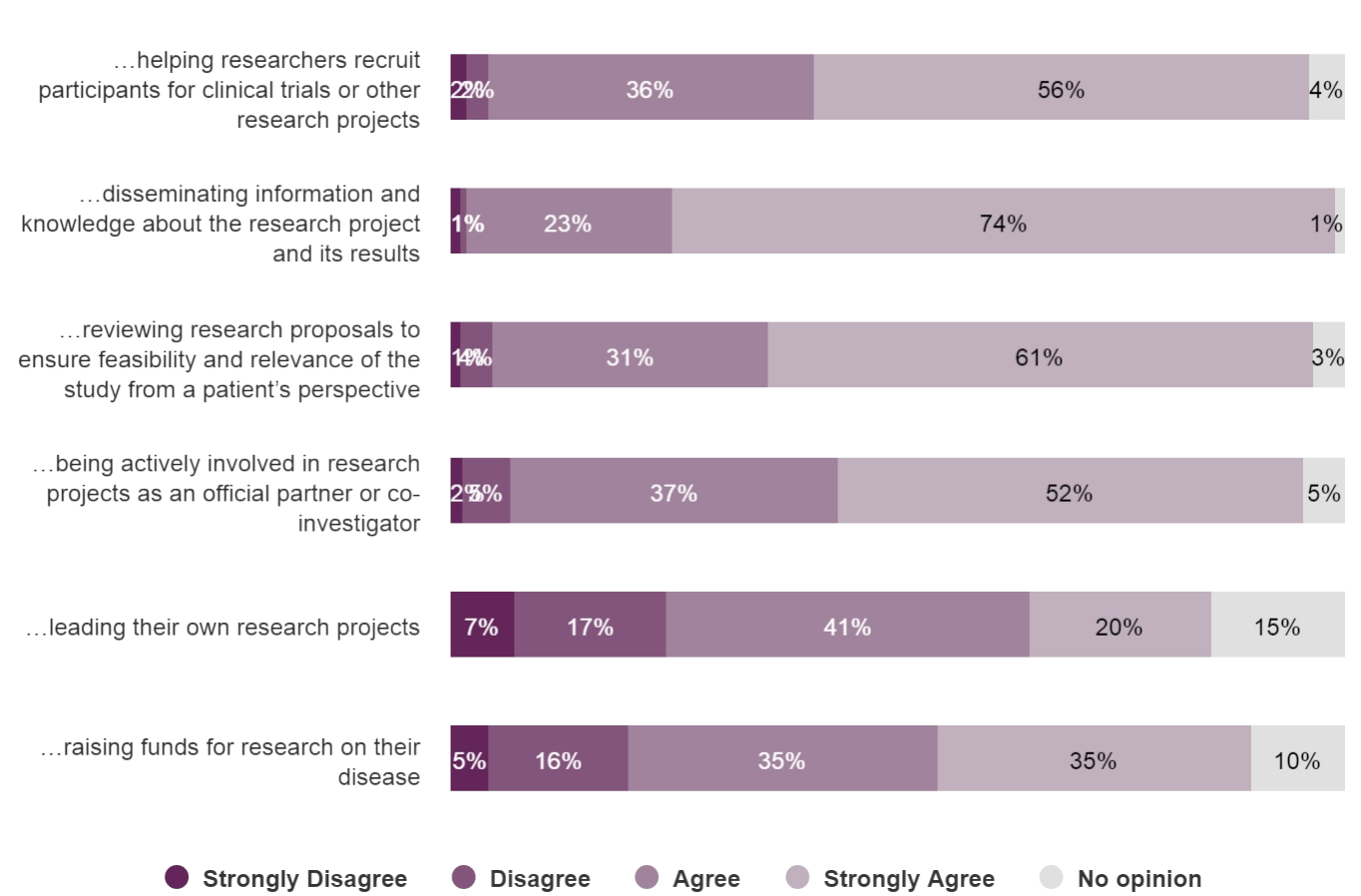
Within the next 10 years, in order to help ease the symptoms of your rare disease, you would need access to:



As a patient representative, do you think that PATIENT ORGANISATIONS should contribute in research for rare diseases by...

	STRON... DISAG...	DISAG...	AGREE	STRON... AGREE	NO OPINION	TOTAL
...helping researchers recruit participants for clinical trials or other research projects	5	6	88	135	9	243
...disseminating information and knowledge about the research project and its results	3	2	56	180	2	243
...reviewing research proposals to ensure feasibility and relevance of the study from a patient's perspective	3	9	75	148	8	243
...being actively involved in research projects as an official partner or co-investigator	4	13	89	126	11	243
...leading their own research projects	18	41	99	49	36	243
...raising funds for research on their disease	11	38	84	85	25	243
TOTAL	44	109	491	723	91	1,458

As a patient representative, do you think that PATIENT ORGANISATIONS should contribute in research for rare diseases by...



In order to be fully involved in research for the rare disease(s) you represent, what type of support would your organisation need most? Several answers possible

	N
Financial support	194
Human resources	168
Knowledge or abilities	147
Other, please specify...	13
I don't know	8
Nothing	2
<b>TOTAL</b>	<b>243</b>

In order to be fully involved in research for the rare disease(s) you represent, what type of support would your organisation need most? Several answers possible

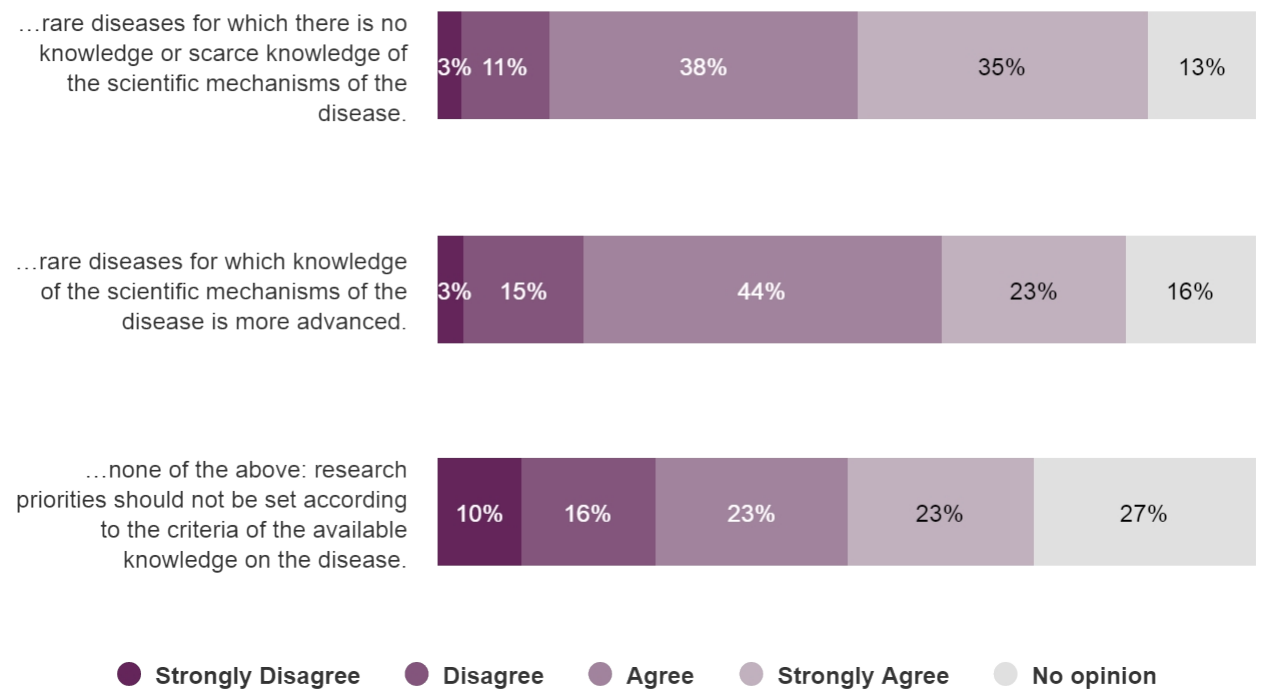




Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on...

	STRON... DISAGR...	DISAGR...	AGREE	STRON... AGREE	NO OPINION	TOTAL
...rare diseases for which there is no knowledge or scarce knowledge of the scientific mechanisms of the disease.	62	204	711	670	246	1,893
...rare diseases for which knowledge of the scientific mechanisms of the disease is more advanced.	64	277	822	423	292	1,878
...none of the above: research priorities should not be set according to the criteria of the available knowledge on the disease.	200	314	450	436	515	1,915
TOTAL	326	795	1,983	1,529	1,053	5,686

Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on...



Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on...

	STRON... DISAGR...	DISAGR...	AGREE	STRON... AGREE	NO OPINION	TOTAL
...rare diseases for which no treatment is available, be it to cure the disease or to manage its symptoms.	57	127	770	835	192	1,981
...rare diseases for which there are already treatments to manage the symptoms but no treatment to cure the disease.	58	182	891	632	218	1,981
...rare diseases for which there are already treatments to stop the progression of the disease but no curative treatment.	65	213	897	551	256	1,982
...none of the above: research priorities should not be set according to the criteria of available treatments.	259	402	392	347	584	1,984
TOTAL	439	924	2,950	2,365	1,250	7,928

Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on...

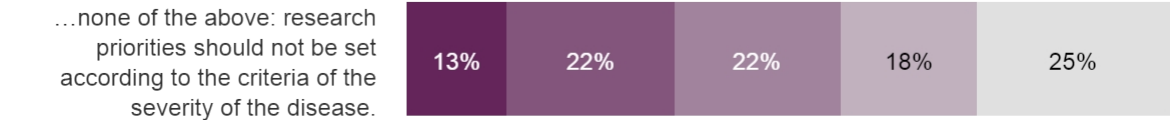


● Strongly Disagree   ● Disagree   ● Agree   ● Strongly Agree   ● No opinion

Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on...

	STRONGLY DISAGREE	DISAGREE	AGREE	STRONGLY AGREE	NO OPINION	TOTAL
...rare diseases for which a treatment (symptomatic or curative) is available but is very expensive, either for the patient (and family) or for society.	58	229	872	489	276	1,924
...rare diseases that have very high associated costs for the patient (and family) or for society (e.g. respite care, adapted living).	55	185	891	514	275	1,920
...none of the above: research priorities should not be set according to the criteria of the severity of the disease.	258	424	421	344	486	1,933
TOTAL	371	838	2,184	1,347	1,037	5,777

Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on...

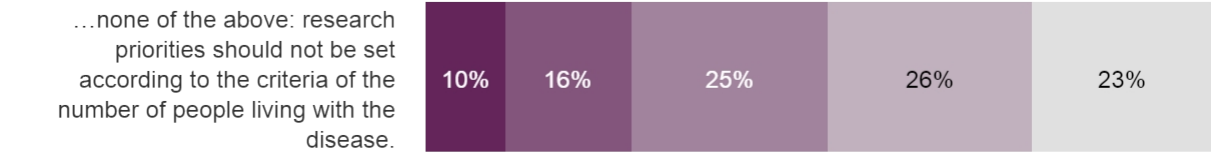


● Strongly Disagree   ● Disagree   ● Agree   ● Strongly Agree   ● No opinion

Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on...

	STRONGLY DISAGREE	DISAGREE	AGREE	STRONGLY AGREE	NO OPINION	TOTAL
...rare diseases that affect very few people and for which raising funds and public awareness is harder.	74	244	782	498	286	1,884
...rare diseases that are more common and where research could benefit more people.	76	289	810	466	237	1,878
...none of the above: research priorities should not be set according to the criteria of the number of people living with the disease.	201	312	483	500	437	1,933
TOTAL	351	845	2,075	1,464	960	5,695

Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on...

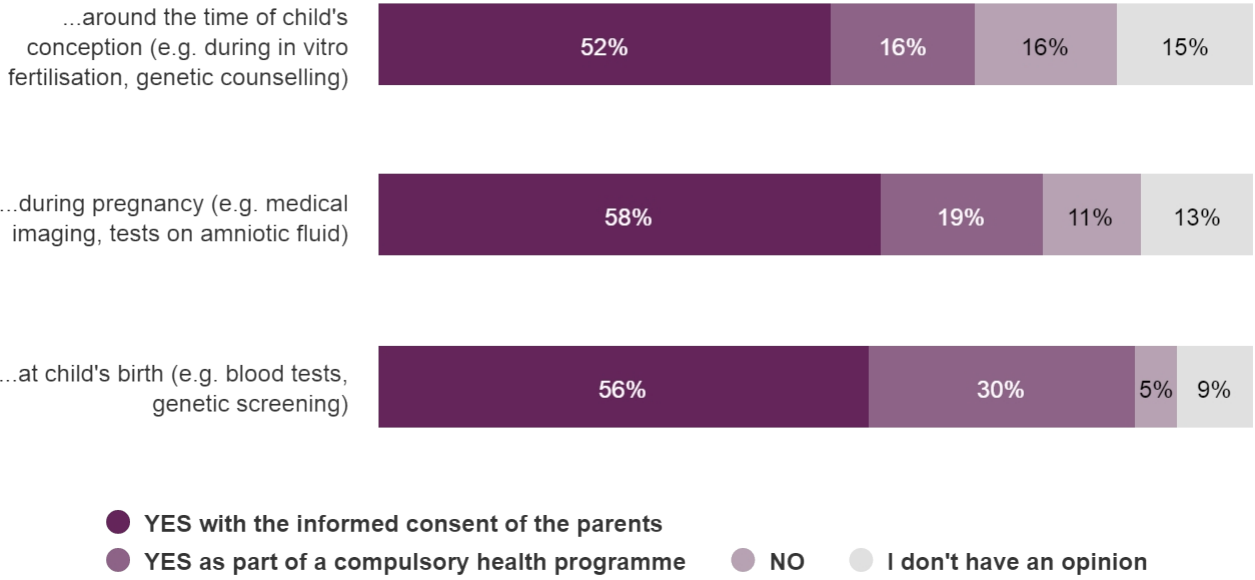


● Strongly Disagree   ● Disagree   ● Agree   ● Strongly Agree   ● No opinion

In your opinion, in order to diagnose rare diseases at an early stage, should tests for rare diseases be performed...

	YES WITH THE INFORMED CONSENT OF THE PARENTS	YES AS PART OF A COMPULS... HEALTH PROGRAM...	NO	I DON'T HAVE AN OPINION	TOTAL
...around the time of child's conception (e.g. during in vitro fertilisation, genetic counselling)	1,903	599	599	561	3,662
...during pregnancy (e.g. medical imaging, tests on amniotic fluid)	2,110	678	414	460	3,662
...at child's birth (e.g. blood tests, genetic screening)	2,062	1,115	171	314	3,662
TOTAL	6,075	2,392	1,184	1,335	10,986

In your opinion, in order to diagnose rare diseases at an early stage, should tests for rare diseases be performed...



In your opinion, for which conditions should newborns be tested as part of a health programme? Several answers possible	
	N
Conditions that COULD BE prevented and/or treated	2,879
Conditions that may affect other family members (inherited conditions)	2,556
Conditions for which appropriate disease management (diet, education...) can improve health and quality of life	2,460
Conditions that may be developed later in life	1,706
Conditions that CANNOT YET BE prevented and/or treated	1,453
I don't have an opinion	218
Other, please specify...	59
None of the above	49
TOTAL	3,647

