*Sample information : **Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

SURVEY RESULTS

The opinion of people living with a rare disease on newborn screening

A Rare Barometer Survey with the Screen4Care research project

Target population :

- People living with a rare disease
- Family members of those living with a rare disease

May 24 - July 23 2023

6,179 respondents worldwide and

5,569 in Europe

24 languages

50 countries

1,331 diseases represented

*Sample information : **Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

DASHBOARD FOR EUROPE

HOW TO USE THIS DASHBOARD

In this dashboard, you will find results for every question of the Rare Barometer survey on the opinion of people living with a rare disease on newborn screening.

Please do not use results of questions for which there are less than 30 respondents.

Please refer to Rare Barometer or add the Rare Barometer logo when using the results.

LANGUAGES

You can change the language at the bottom left of this page, and have access to the questions and modalities as they appeared to respondents in the 24 languages of the survey.

Translation is not available for new variables that were calculated after the questionnaire was closed and for some comments added in this dashboard.

INFORMATION

For more information:

- contact the Rare Barometer team at rare.barometer@eurordis.org
- visit the Rare Barometer website at eurordis.org/voices
- visit our newborn screening-dedicated webpage: eurordis.org/rare-barometer-survey

LIST OF CONTENT

MORE ON THE SURVEY

Research questions and in-depth analysis of European results are in the **full report in English**: http://tiny.cc/RB_NBS

Key results are available in the **factsheets**: http://tiny.cc/RB_NBS

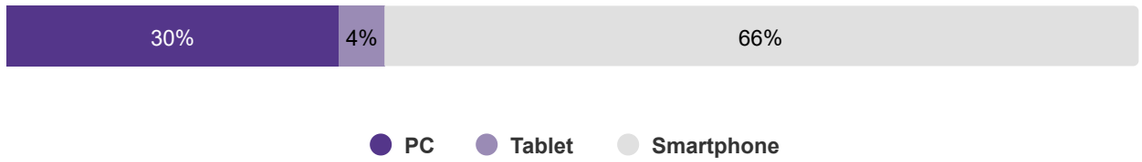
Original questionnaire: tiny.cc/RB_NBS_questionnaire

*Sample information : **Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

Number of respondents

5,569

Device used to answser



Average number of minutes to fill the questionnaire

26

Mean

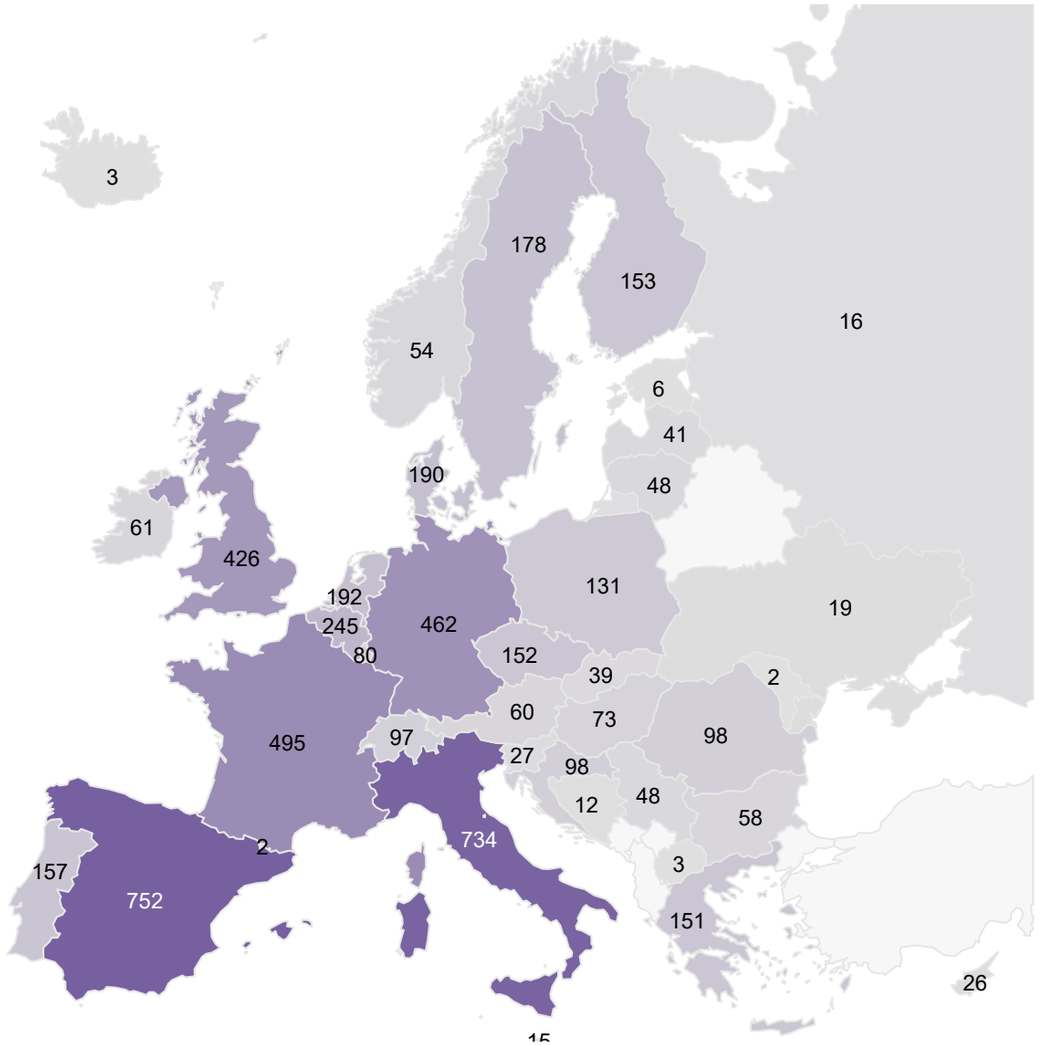
Median number of minutes to fill the questionnaire

19

Median

Sample information : ***Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

In which country do you live?



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Sample size 5569 responses

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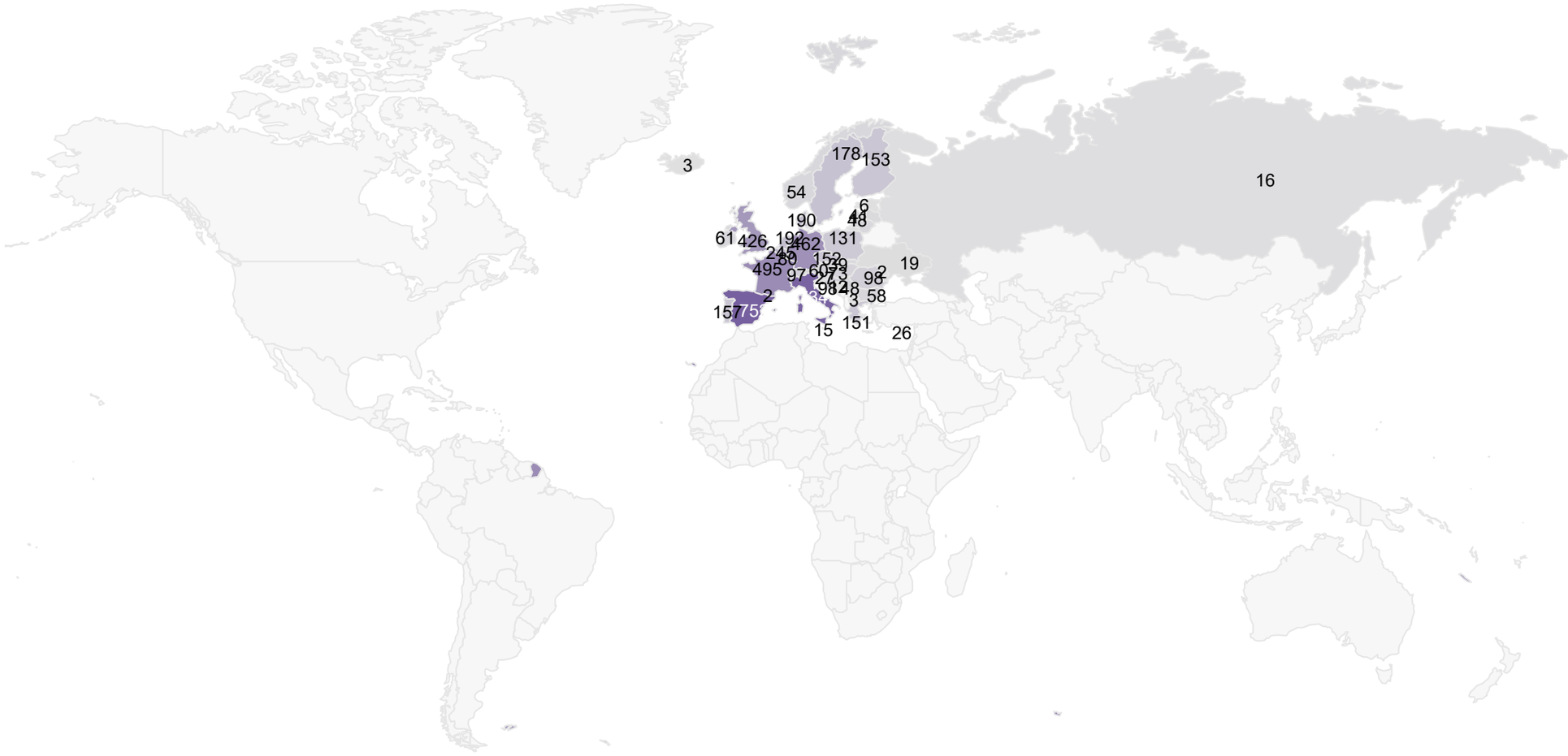

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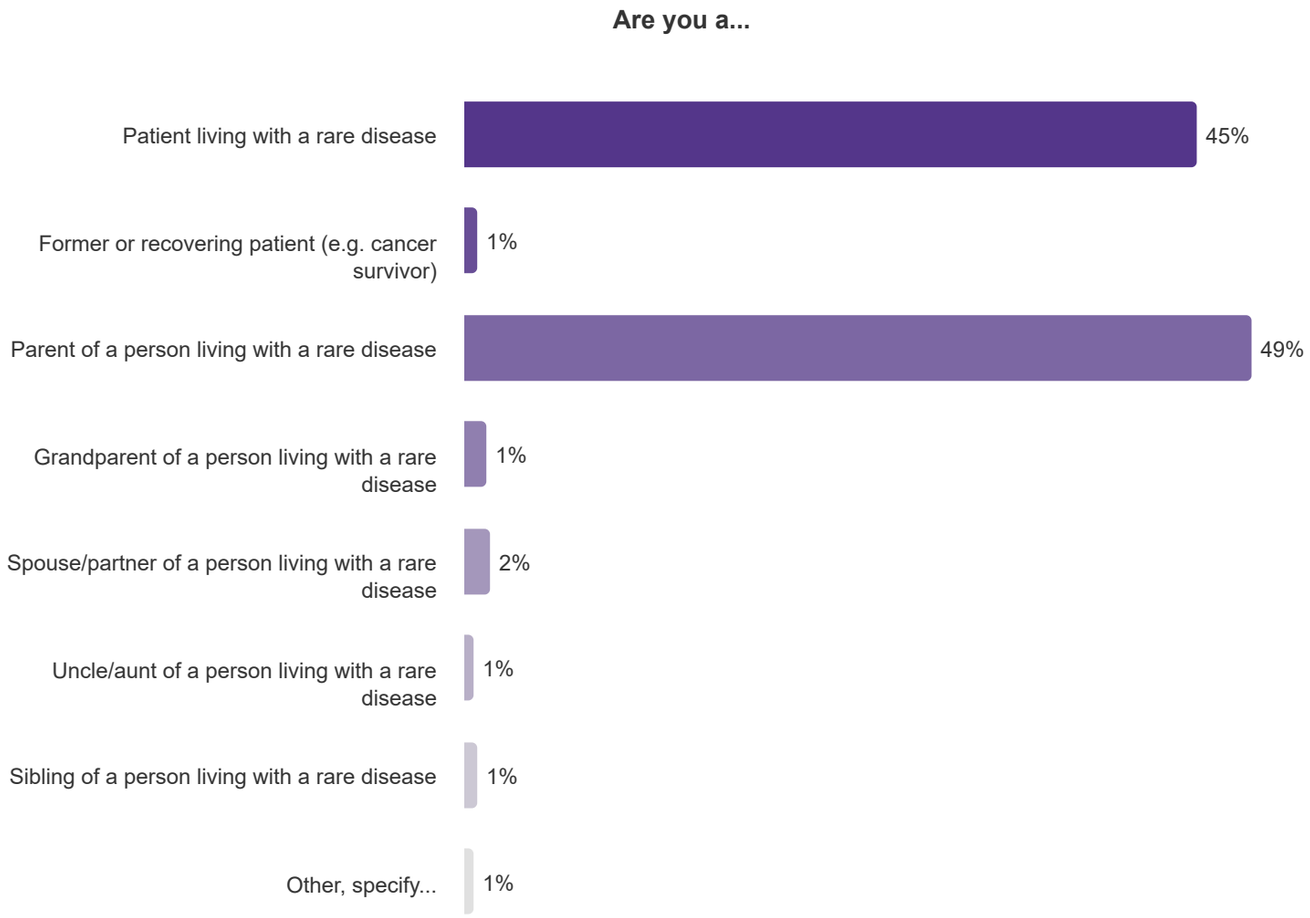
Sample size 5569 responses

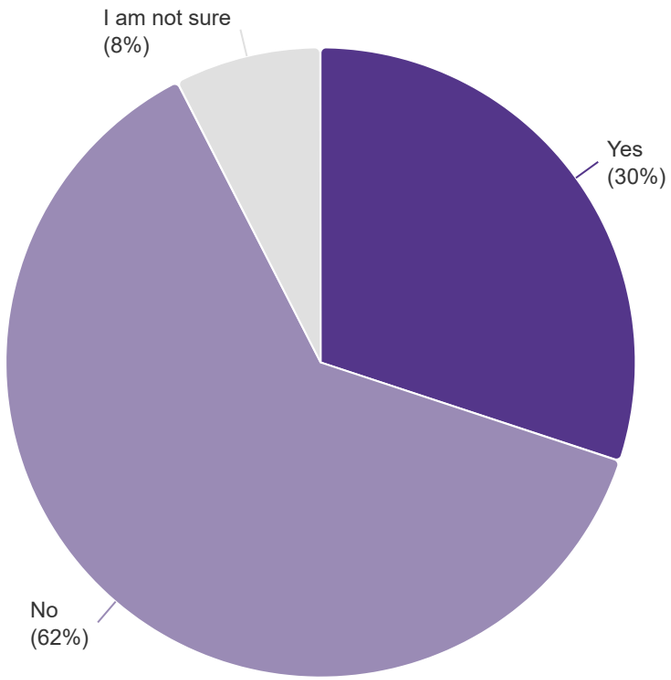


Are you a...

	N
Patient living with a rare disease	2,514
Former or recovering patient (e.g. cancer survivor)	53
Parent of a person living with a rare disease	2,701
Grandparent of a person living with a rare disease	80
Spouse/partner of a person living with a rare disease	93
Uncle/aunt of a person living with a rare disease	39
Sibling of a person living with a rare disease	49
Other, specify...	40
TOTAL	**5,569**

Sample information : ***Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

Are you also a patient representative, i.e., involved in voluntary and/or policy activities to support the cause of rare diseases?

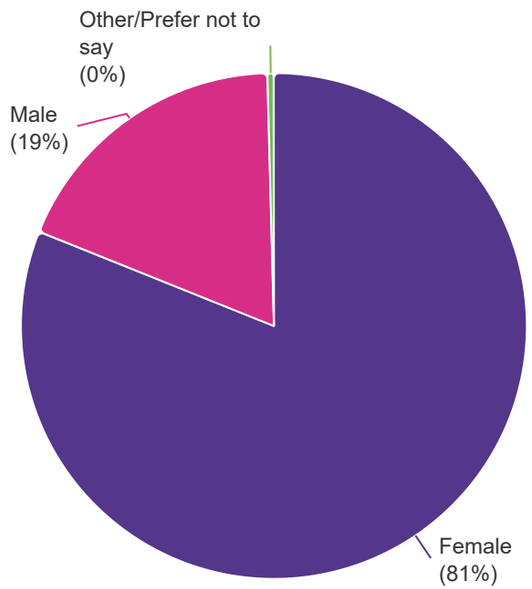


Are you also a patient representative, i.e., involved in voluntary and/or policy activities to support the cause of rare diseases?

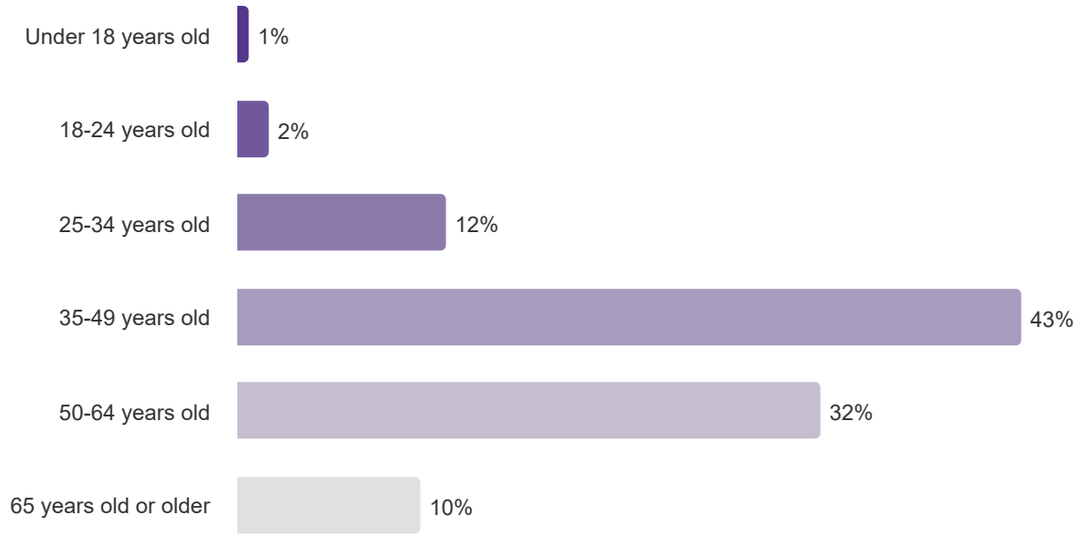
	N
Yes	1,675
No	3,475
I am not sure	419
TOTAL	**5,569**

*Sample information : **Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

Are you:



How old are you?



Are you:

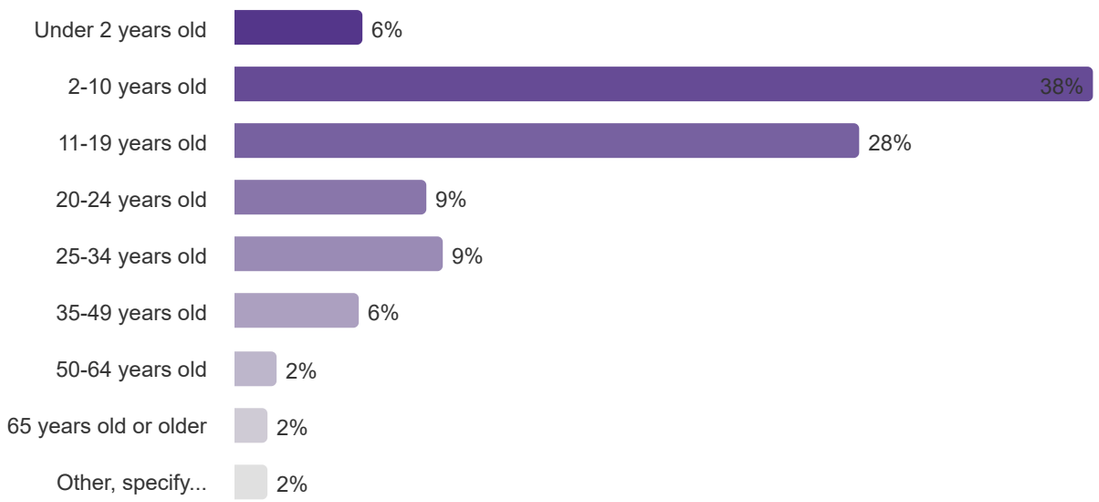
	N
Female	4,235
Male	967
Other/Prefer not to say	21
TOTAL	**5,223**

How old are you?

	N
Under 18 years old	35
18-24 years old	92
25-34 years old	590
35-49 years old	2,206
50-64 years old	1,640
65 years old or older	518
TOTAL	**5,081**

*Sample information : **Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

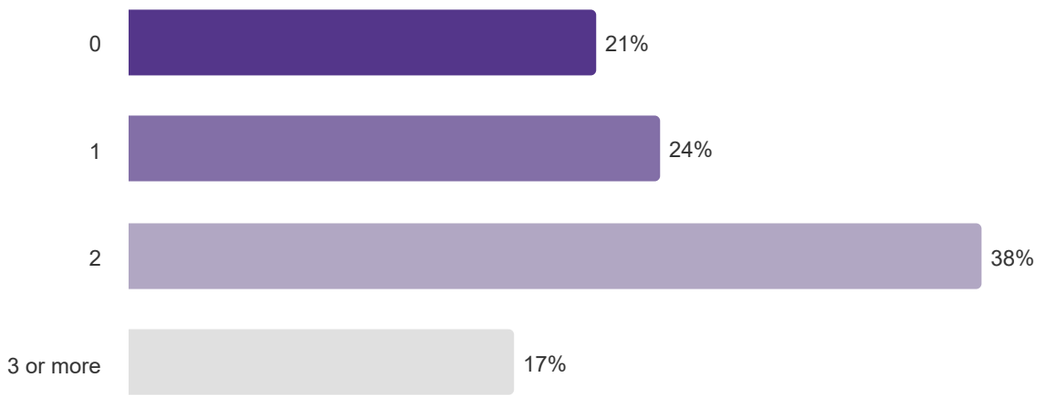
How old is the person affected by the rare disease?



How old is the person affected by the rare disease?

	N
Under 2 years old	157
2-10 years old	1,040
11-19 years old	758
20-24 years old	235
25-34 years old	255
35-49 years old	152
50-64 years old	53
65 years old or older	41
Other, specify...	42
TOTAL	**2,733**

How many children do you have?

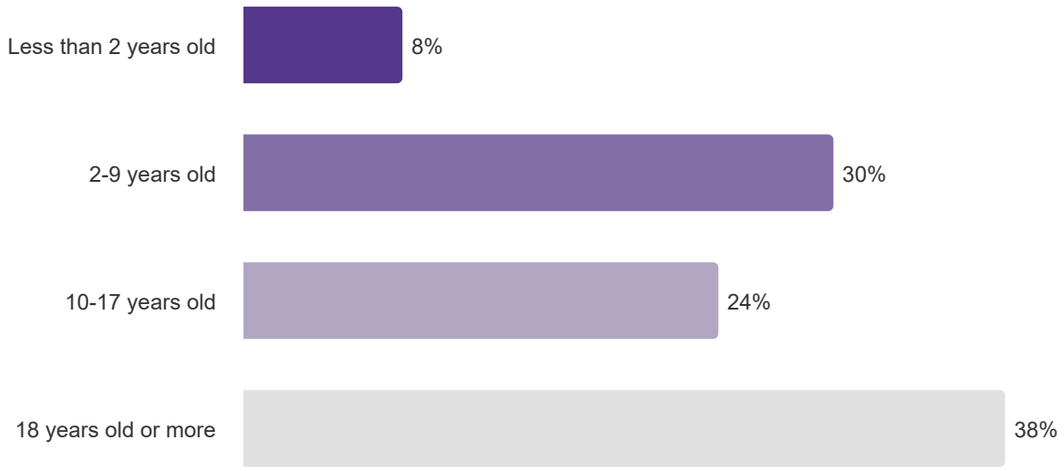


How many children do you have?

	N
	1,059
1	1,203
2	1,932
3 or more	876
TOTAL	**5,070**

*Sample information : **Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

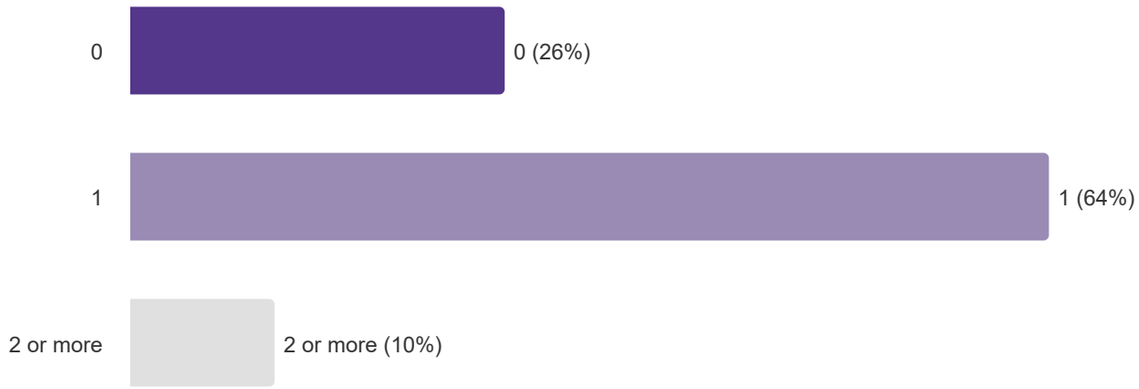
How old is your youngest child?



How old is your youngest child?

	N
Less than 2 years old	324
2-9 years old	1,189
10-17 years old	959
18 years old or more	1,538
TOTAL	**4,010**

How many children affected by a rare disease do you have?

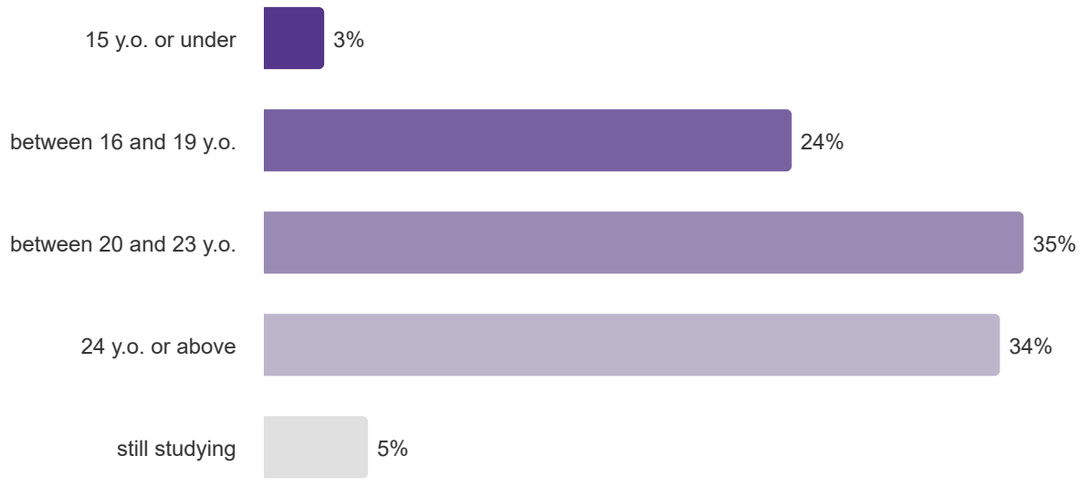


How many children affected by a rare disease do you have?

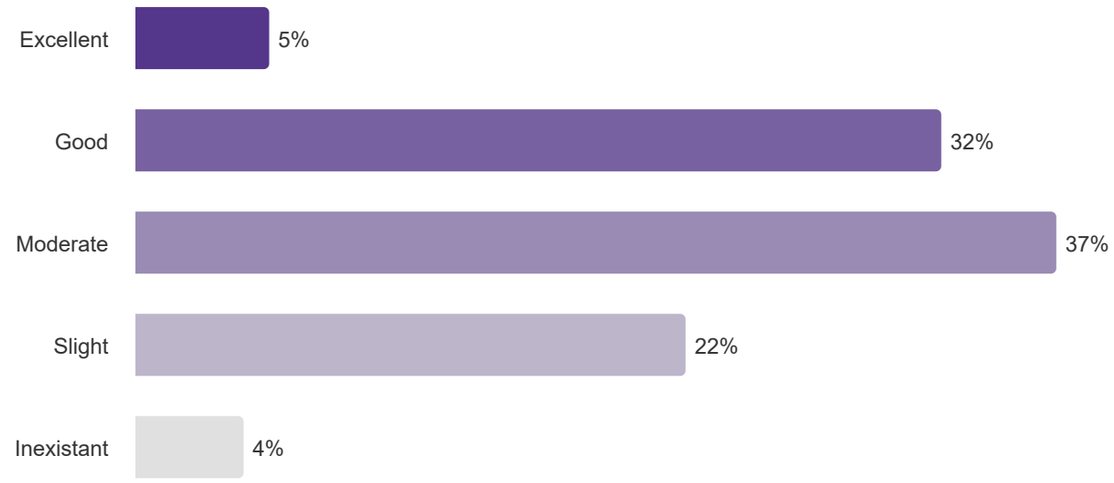
	N
	1,045
1	2,560
2 or more	405
TOTAL	**4,010**

*Sample information : **Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

How old were you when you stopped full-time education?



How would you describe your knowledge of genetics?



How old were you when you stopped full-time education?

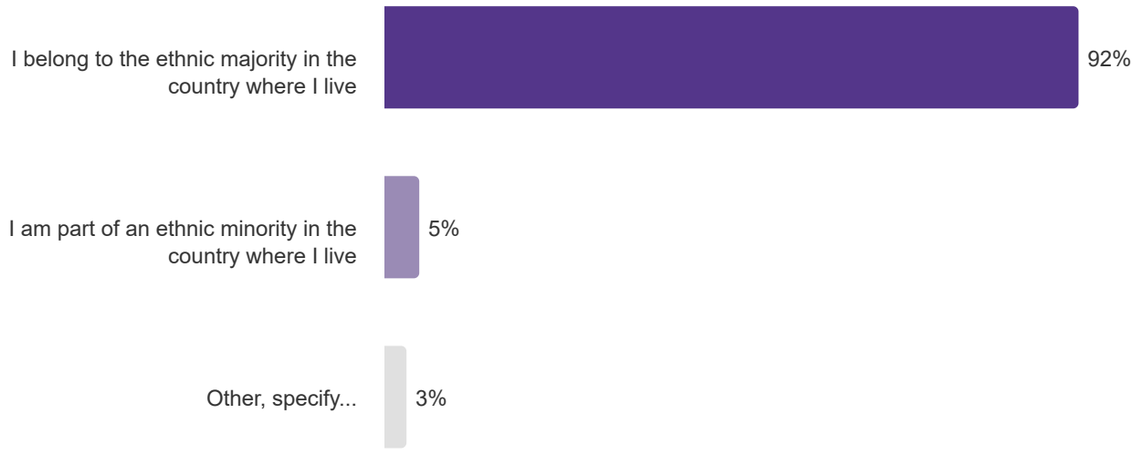
	N
15 y.o. or under	142
between 16 and 19 y.o.	1,222
between 20 and 23 y.o.	1,757
24 y.o. or above	1,699
still studying	243
TOTAL	**5,063**

How would you describe your knowledge of genetics?

	N
Excellent	270
Good	1,618
Moderate	1,850
Slight	1,105
Inexistant	220
TOTAL	**5,063**

Sample information : ***Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

How would you best describe yourself?



How would you best describe yourself?

	N
I belong to the ethnic majority in the country where I live	4,217
I am part of an ethnic minority in the country where I live	224
Other, specify...	139
TOTAL	**4,580**

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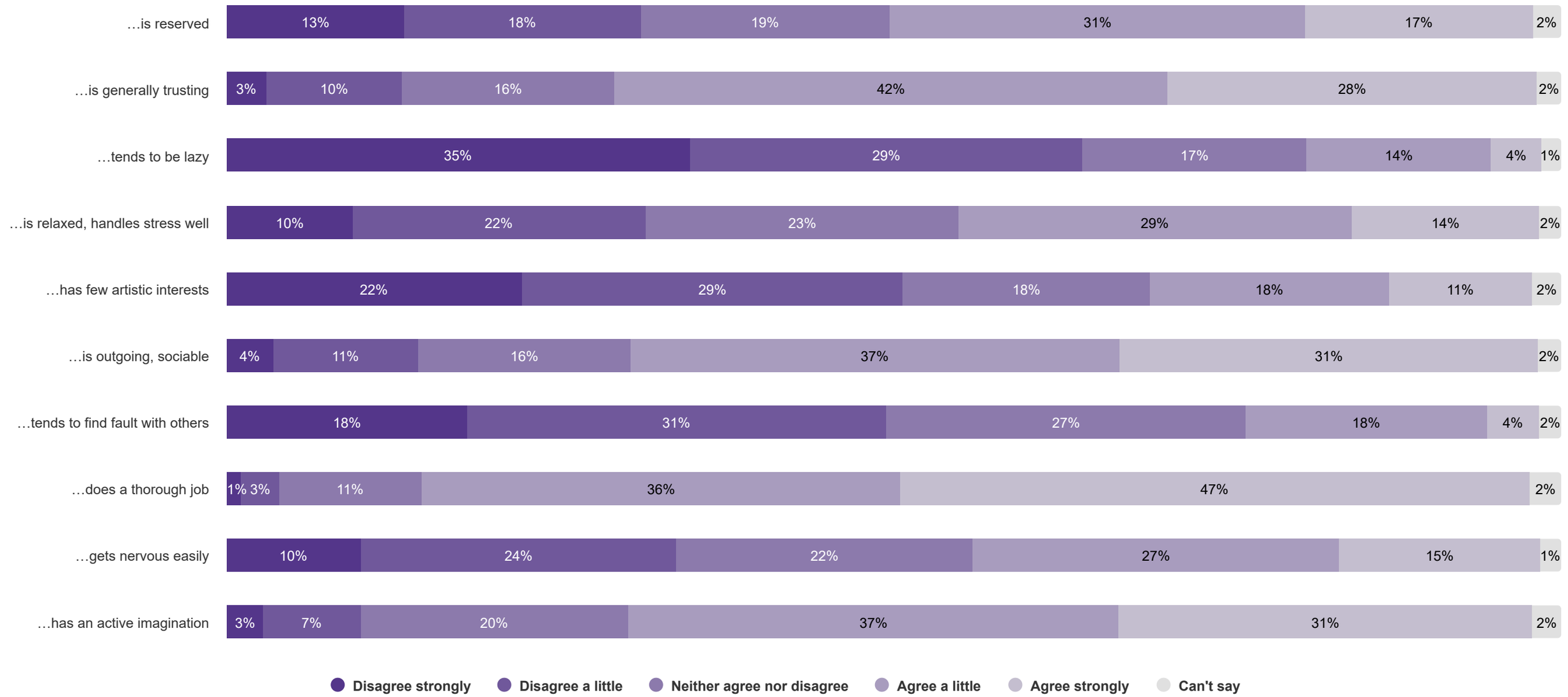
Sample size 5569 responses

More questions about you... This information will allow us to analyse the data in more depth. I see myself as someone who:



More questions about you... This information will allow us to analyse the data in more depth. I see myself as someone who:

	DISAGREE STRONGLY	DISAGREE A LITTLE	NEITHER AGREE NOR DISAGREE	AGREE A LITTLE	AGREE STRONGLY	CAN'T SAY	TOTAL
…is reserved	680	905	946	1,585	867	101	**5,084**
…is generally trusting	156	514	811	2,110	1,406	87	**5,084**
…tends to be lazy	1,769	1,495	857	699	193	71	**5,084**
…is relaxed, handles stress well	483	1,119	1,193	1,496	712	81	**5,084**
…has few artistic interests	1,132	1,449	940	913	544	106	**5,084**
…is outgoing, sociable	183	551	810	1,862	1,594	84	**5,084**
…tends to find fault with others	922	1,595	1,368	921	196	81	**5,083**
…does a thorough job	56	150	542	1,823	2,396	116	**5,083**
…gets nervous easily	514	1,202	1,129	1,397	765	76	**5,083**
…has an active imagination	144	370	1,020	1,868	1,576	105	**5,083**

Sample information : ***Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

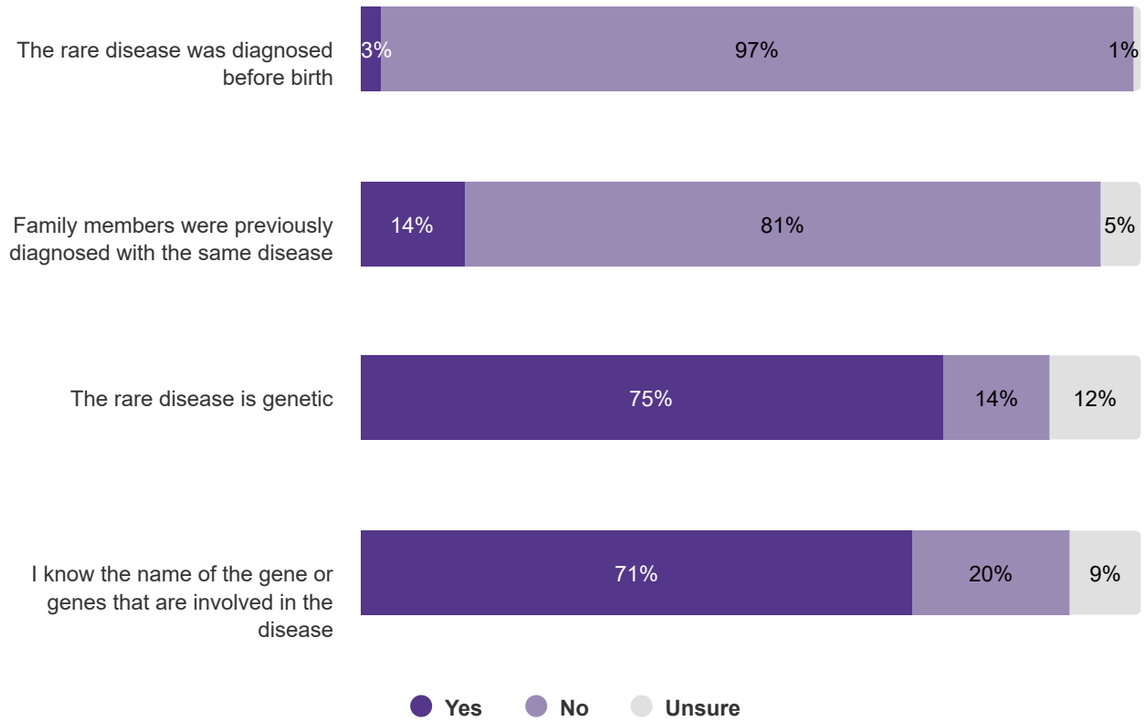
Please select the sentence that best describes your situation:

Please select the sentence that best describes your situation:

	N
I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test)	4,984
I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests	258
I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease	110
I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED	195
Other, specify...	22
TOTAL	**5,569**

Note: These questions were only asked to respondents who said that the rare disease has been diagnosed. The last item was only asked to those who indicated that the rare diseases is genetic.

Do the following sentences apply to your situation?



Do the following sentences apply to your situation?

	YES	NO	UNSURE	TOTAL
The rare disease was diagnosed before birth	149	5,139	32	**5,320**
Family members were previously diagnosed with the same disease	725	4,329	266	**5,320**
The rare disease is genetic	3,981	725	614	**5,320**
I know the name of the gene or genes that are involved in the disease	3,261	927	406	**4,594**

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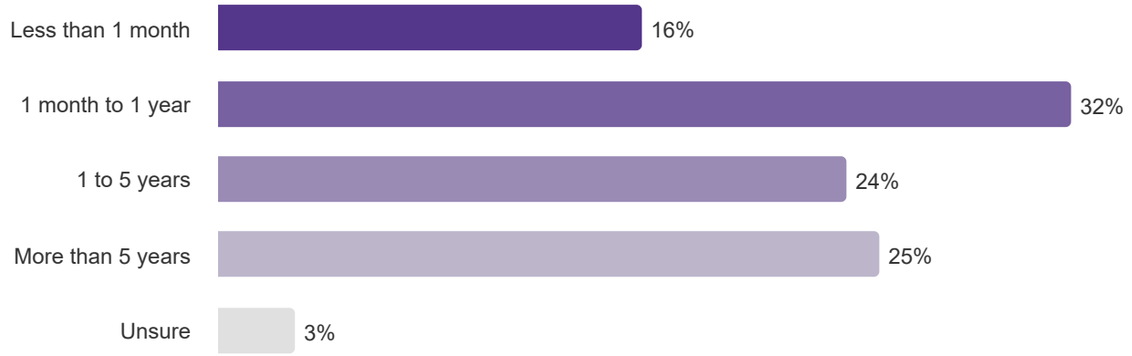
Sample size 5569 responses

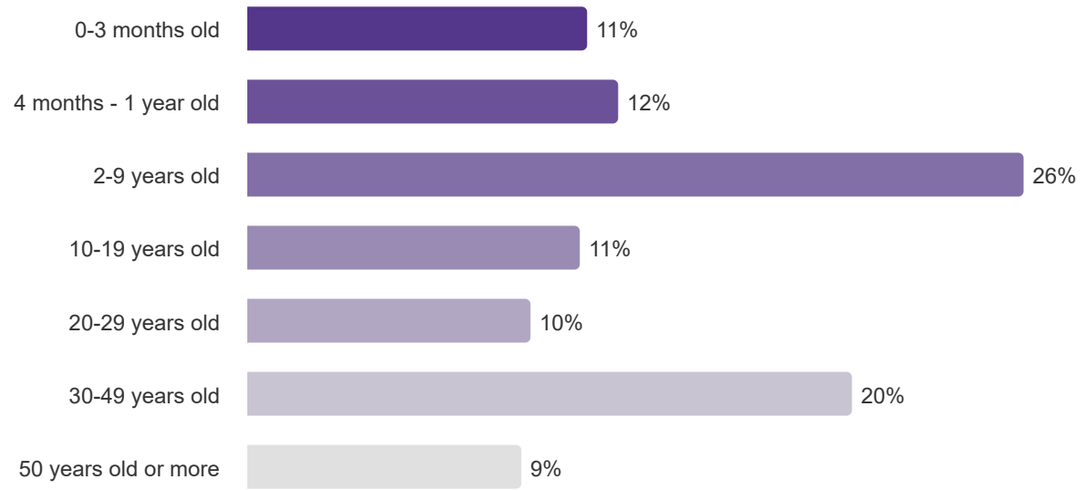
How long did it take from the first medical encounter for the diagnosis to be confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g., biopsy, blood or urine test):



How long did it take from the first medical encounter for the diagnosis to be confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g., biopsy, blood or urine test):

	N
Less than 1 month	825
1 month to 1 year	1,655
1 to 5 years	1,222
More than 5 years	1,284
Unsure	152
TOTAL	**5,138**

How old were you when you received a confirmed diagnosis?



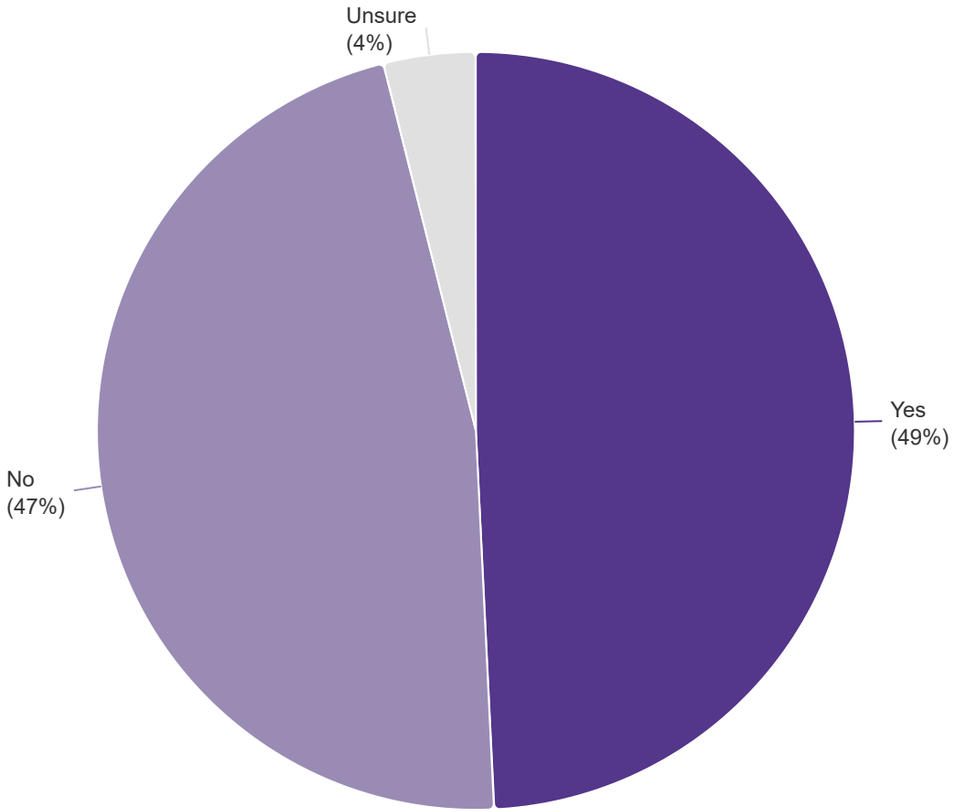
How old were you when you received a confirmed diagnosis?

	N
0-3 months old	587
4 months - 1 year old	638
2-9 years old	1,335
10-19 years old	574
20-29 years old	489
30-49 years old	1,040
50 years old or more	474
TOTAL	**5,137**

Note: These questions were only asked to respondents who said that the rare disease has been diagnosed.

*Sample information : **Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

Was the rare disease diagnosed through standard tests carried out at birth?

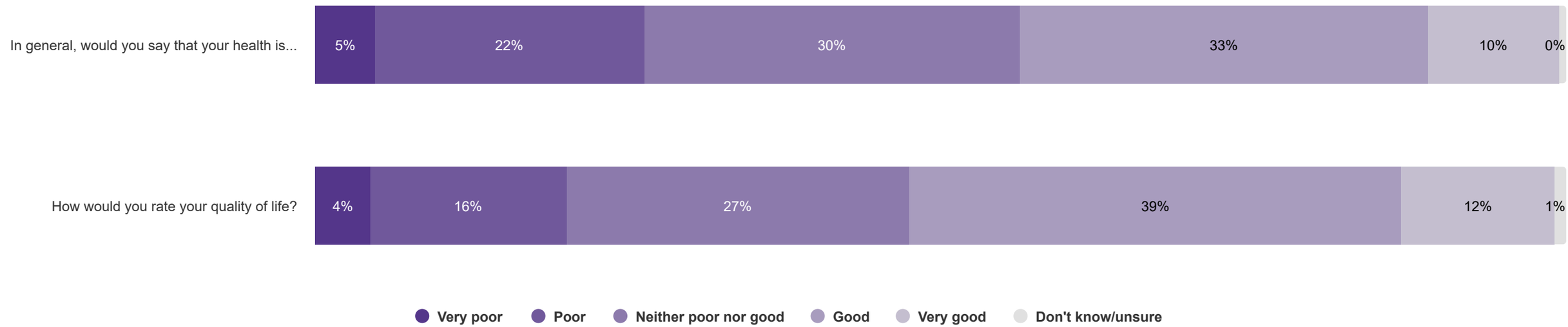


This question was only asked when patients were diagnosed under 3 months old.

Was the rare disease diagnosed through standard tests carried out at birth?

	N
Yes	289
No	275
Unsure	23
TOTAL	**587**

Sample information : ***Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses



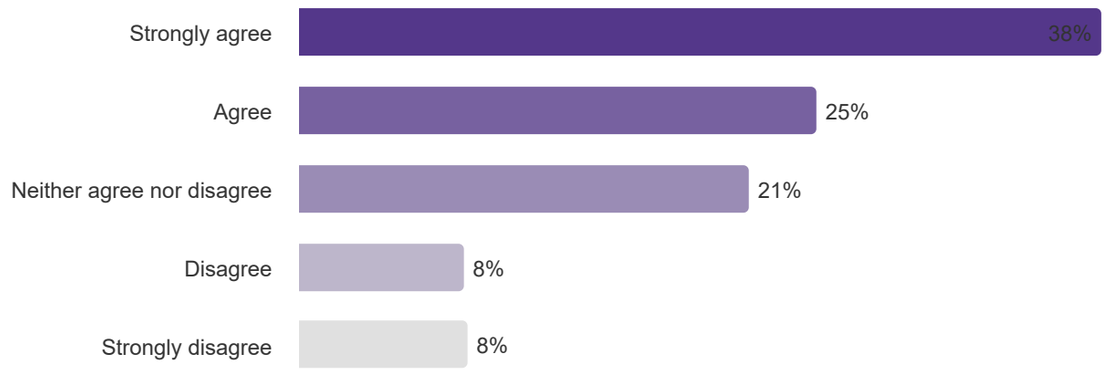
	VERY POOR	POOR	NEITHER POOR NOR GOOD	GOOD	VERY GOOD	DON'T KNOW/UNSURE	TOTAL
In general, would you say that your health is...	274	1,200	1,667	1,822	579	27	**5,569**
How would you rate your quality of life?	250	875	1,524	2,191	683	46	**5,569**

*Sample information : **Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

…I would have liked to be diagnosed AT BIRTH

	N
Strongly agree	965
Agree	624
Neither agree nor disagree	542
Disagree	199
Strongly disagree	204
TOTAL	**2,534**

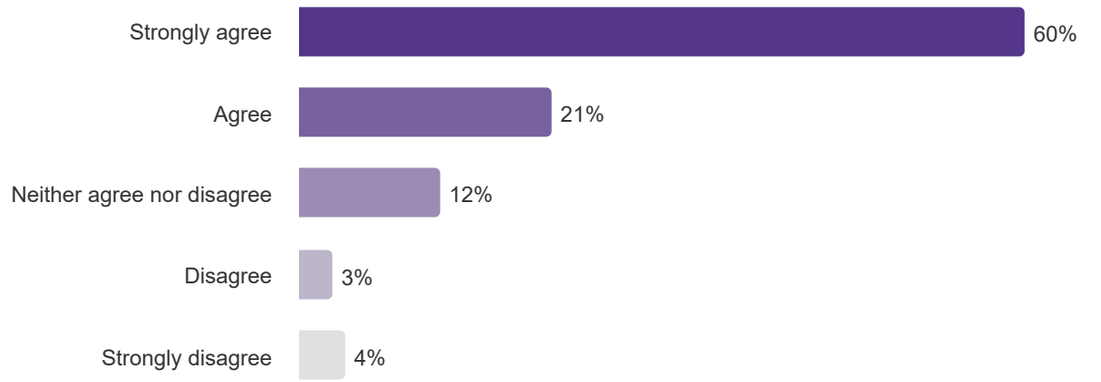
…I would have liked to be diagnosed AT BIRTH



…I would have liked the person I care for to be diagnosed AT BIRTH

	N
Strongly agree	1,812
Agree	634
Neither agree nor disagree	354
Disagree	86
Strongly disagree	116
TOTAL	**3,002**

…I would have liked the person I care for to be diagnosed AT BIRTH



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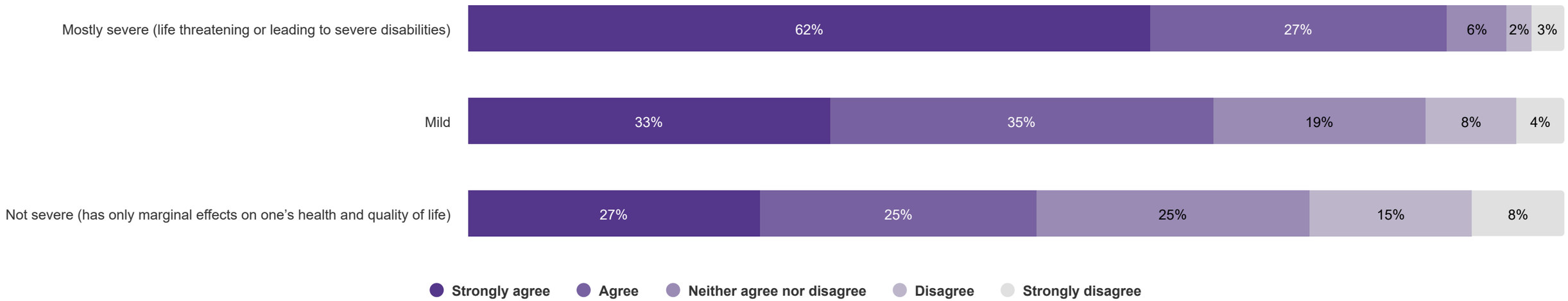
Sample size 5569 responses

In your opinion, should ANY RARE DISEASE be screened at birth, PROVIDED THAT it is:

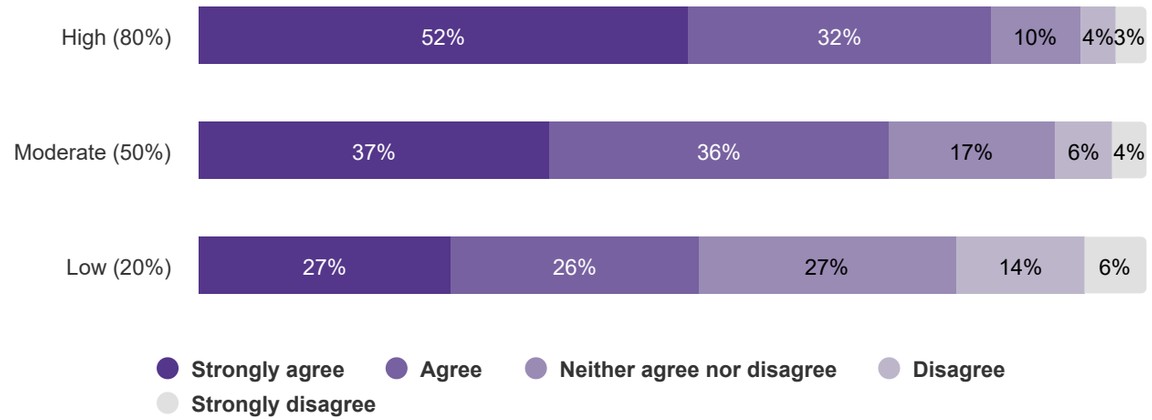


In your opinion, should ANY RARE DISEASE be screened at birth, PROVIDED THAT it is:

	STRONGLY AGREE	AGREE	NEITHER AGREE NOR DISAGREE	DISAGREE	STRONGLY DISAGREE	TOTAL
Mostly severe (life threatening or leading to severe disabilities)	3,475	1,502	308	126	158	**5,569**
Mild	1,844	1,953	1,076	459	237	**5,569**
Not severe (has only marginal effects on one's health and quality of life)	1,490	1,407	1,386	824	462	**5,569**

Sample information : ***Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

When screening for a disease, there can be a chance that the disease will not develop even if the test is positive. In your opinion, should ANY rare disease be screened at birth PROVIDED THAT if the test is positive, the chance for the disease to actually appear is:

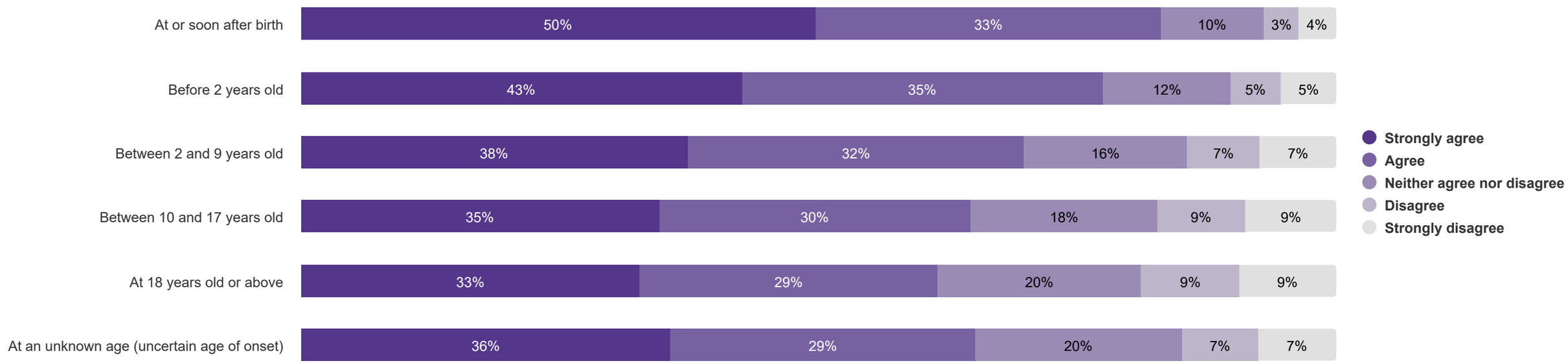


When screening for a disease, there can be a chance that the disease will not develop even if the test is positive. In your opinion, should ANY rare disease be screened at birth PROVIDED THAT if the test is positive, the chance for the disease to actually appear is:

	AGREE	STRONGLY AGREE	NEITHER AGREE NOR DISAGREE	DISAGREE	STRONGLY DISAGREE	TOTAL
High (80%)	1,779	2,886	532	196	176	**5,569**
Moderate (50%)	2,005	2,067	972	329	196	**5,569**
Low (20%)	1,454	1,495	1,514	757	349	**5,569**

*Sample information : **Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

A disease can be diagnosed at birth but only manifest later in life. In your opinion, should ANY rare disease be screened at birth, PROVIDED THAT the first symptoms typically appear:

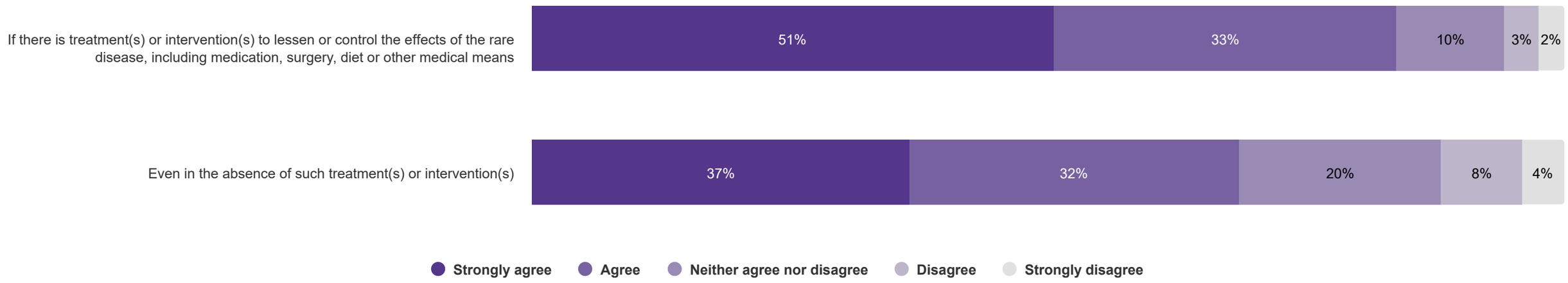


A disease can be diagnosed at birth but only manifest later in life. In your opinion, should ANY rare disease be screened at birth, PROVIDED THAT the first symptoms typically appear:

	STRONGLY AGREE	AGREE	NEITHER AGREE NOR DISAGREE	DISAGREE	STRONGLY DISAGREE	TOTAL
At or soon after birth	2,778	1,854	552	190	195	**5,569**
Before 2 years old	2,380	1,944	685	268	292	**5,569**
Between 2 and 9 years old	2,090	1,808	873	392	406	**5,569**
Between 10 and 17 years old	1,934	1,672	1,006	474	483	**5,569**
At 18 years old or above	1,826	1,603	1,095	529	516	**5,569**
At an unknown age (uncertain age of onset)	1,995	1,637	1,115	406	416	**5,569**

Sample information : ***Cnsqcs__undermined_tests_system is not among "Non-response" And CONTINENT among "Europe" And case_controle does not contain "control" And Consent_rep_leg is not among "No"***
Sample size 5569 responses

The next few questions will be about your opinion on using tests to screen for ANY RARE DISEASE at birth. In your opinion, should ANY RARE DISEASE be screened at birth:



The next few questions will be about your opinion on using tests to screen for ANY RARE DISEASE at birth. In your opinion, should ANY RARE DISEASE be screened at birth:

	STRONGLY AGREE	AGREE	NEITHER AGREE NOR DISAGREE	DISAGREE	STRONGLY DISAGREE	TOTAL
If there is treatment(s) or intervention(s) to lessen or control the effects of the rare disease, including medication, surgery, diet or other medical means	2,825	1,846	584	178	136	**5,569**
Even in the absence of such treatment(s) or intervention(s)	2,047	1,776	1,090	436	220	**5,569**

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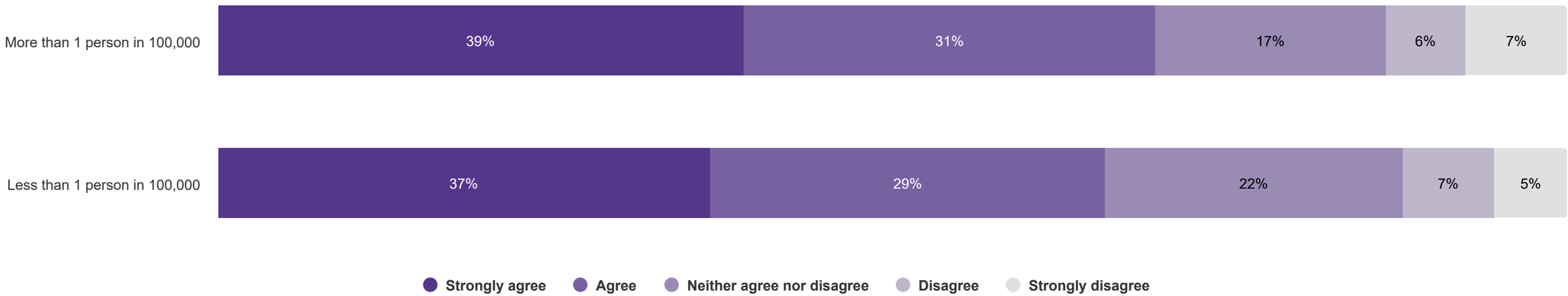
Sample size 5569 responses

In your opinion, should ANY RARE DISEASE be screened at birth PROVIDED THAT it affects:



In your opinion, should ANY RARE DISEASE be screened at birth PROVIDED THAT it affects:

	STRONGLY AGREE	AGREE	NEITHER AGREE NOR DISAGREE	DISAGREE	STRONGLY DISAGREE	TOTAL
More than 1 person in 100,000	2,172	1,703	953	328	413	**5,569**
Less than 1 person in 100,000	2,036	1,631	1,232	375	295	**5,569**

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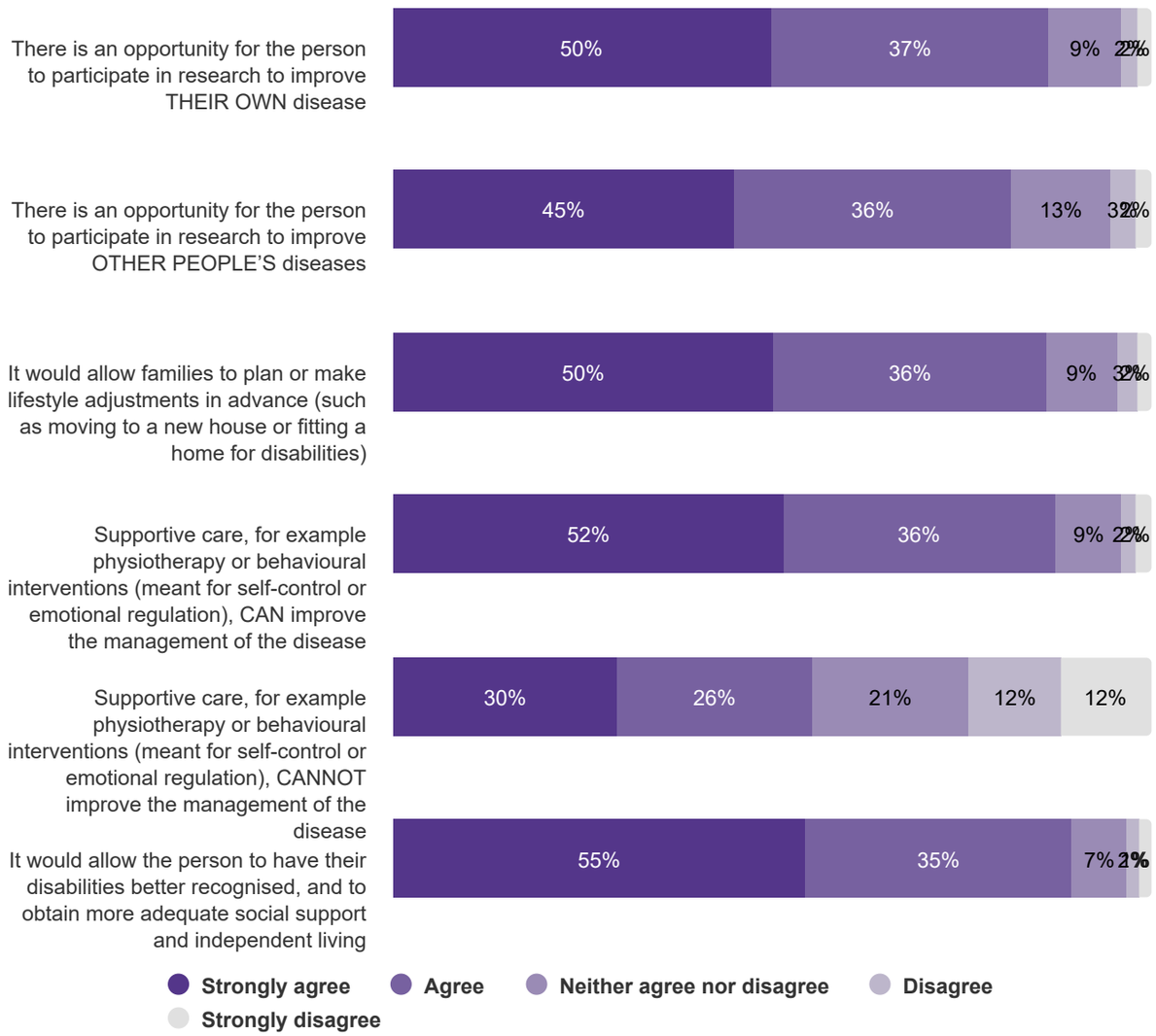
Sample size 5569 responses

In your opinion, should ANY RARE DISEASE be screened at birth IF NO TREATMENT EXISTS AND:



In your opinion, should ANY RARE DISEASE be screened at birth IF NO TREATMENT EXISTS AND:

	STRON… AGREE	AGREE	NEITHER AGREE NOR DISAG…	DISAG…	STRON… DISAG…	TOTAL
There is an opportunity for the person to participate in research to improve THEIR OWN disease	2,792	2,040	527	123	87	**5,569**
There is an opportunity for the person to participate in research to improve OTHER PEOPLE'S diseases	2,521	2,030	731	186	101	**5,569**
It would allow families to plan or make lifestyle adjustments in advance (such as moving to a new house or fitting a home for disabilities)	2,809	2,004	515	153	88	**5,569**
Supportive care, for example physiotherapy or behavioural interventions (meant for self-control or emotional regulation), CAN improve the management of the disease	2,877	2,006	475	112	99	**5,569**
Supportive care, for example physiotherapy or behavioural interventions (meant for self-control or emotional regulation), CANNOT improve the management of the disease	1,656	1,430	1,155	671	657	**5,569**
It would allow the person to have their disabilities better recognised, and to obtain more adequate social support	3,045	1,952	397	101	74	**5,569**

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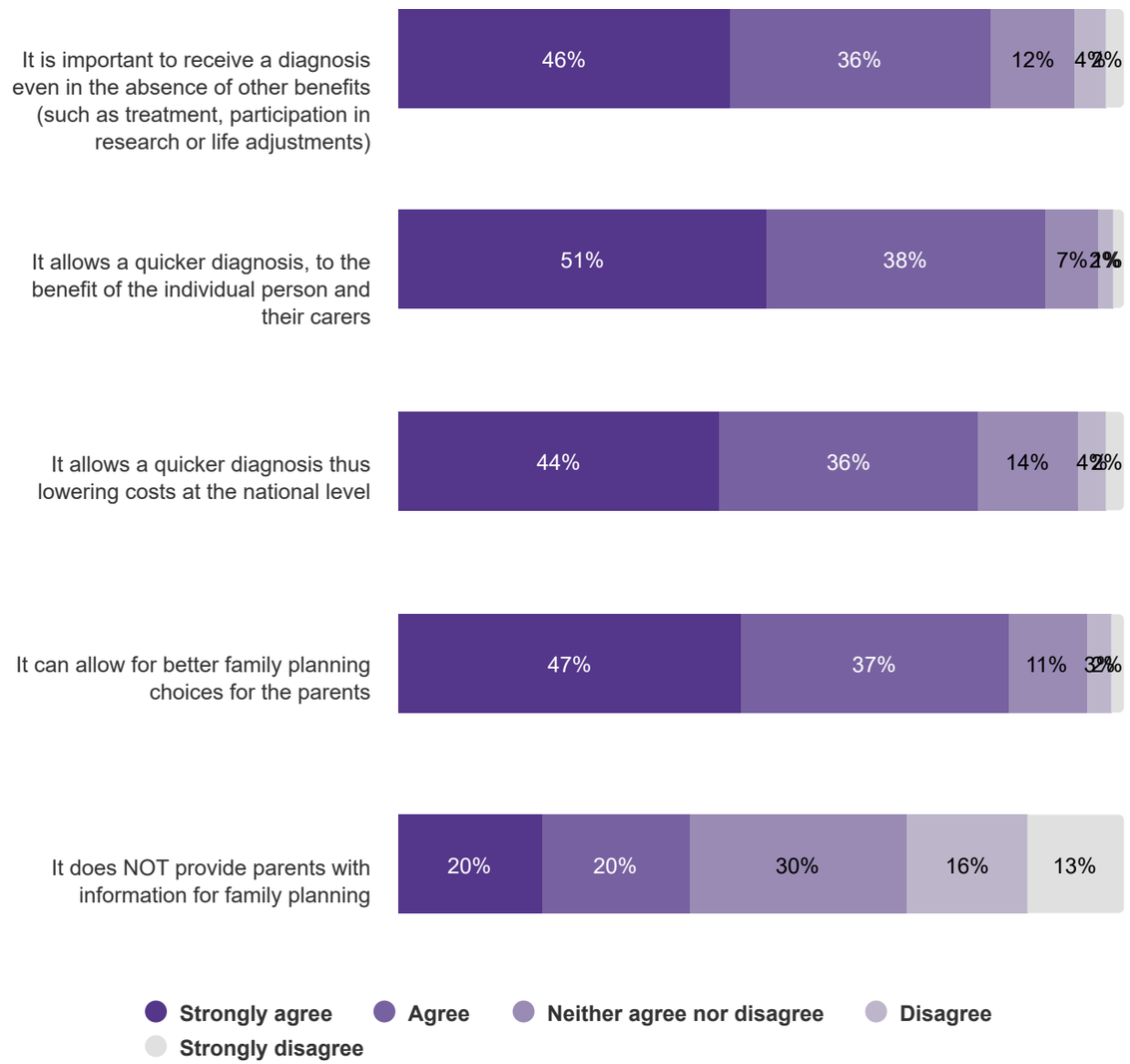

In your opinion, should ANY RARE DISEASE be screened at birth IF NO TREATMENT EXISTS AND:

	STRON... AGREE	AGREE	NEITHER AGREE NOR DISAGREE	DISAGREE	STRON... DISAGREE	TOTAL
It is important to receive a diagnosis even in the absence of other benefits (such as treatment, participation in research or life adjustments)	2,565	1,998	646	239	121	**5,569**
It allows a quicker diagnosis, to the benefit of the individual person and their carers	2,845	2,141	397	116	70	**5,569**
It allows a quicker diagnosis thus lowering costs at the national level	2,471	1,991	776	201	130	**5,569**
It can allow for better family planning choices for the parents	2,641	2,062	606	171	89	**5,569**
It does NOT provide parents with information for family planning	1,117	1,135	1,672	912	733	**5,569**

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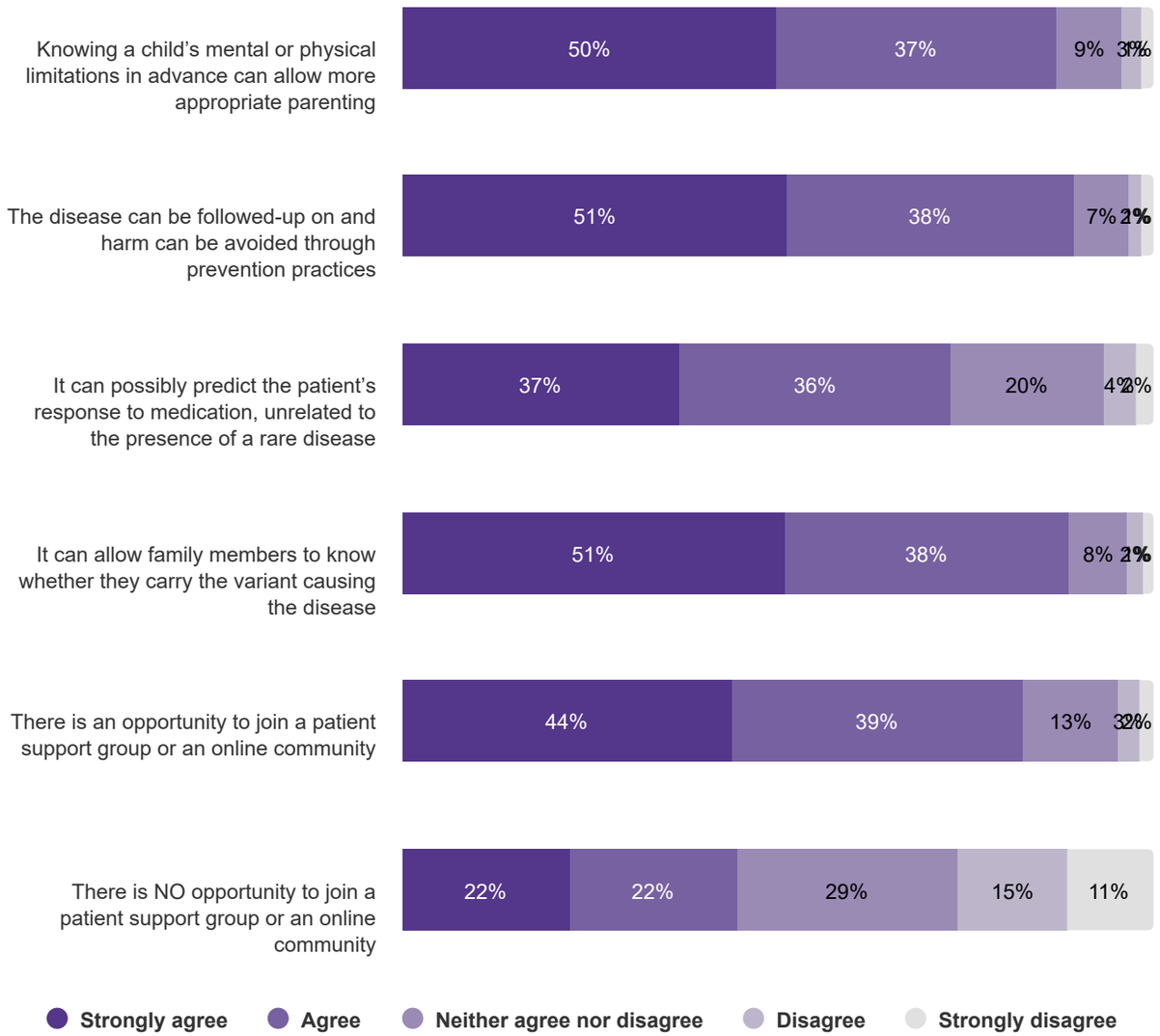

In your opinion, should ANY RARE DISEASE be screened at birth IF NO TREATMENT EXISTS AND:

	STRON... AGREE	AGREE	NEITHER AGREE NOR DISAG...	DISAG...	STRON... DISAG...	TOTAL
Knowing a child's mental or physical limitations in advance can allow more appropriate parenting	2,787	2,080	476	154	72	**5,569**
The disease can be followed-up on and harm can be avoided through prevention practices	2,858	2,136	411	91	73	**5,569**
It can possibly predict the patient's response to medication, unrelated to the presence of a rare disease	2,068	2,016	1,132	237	116	**5,569**
It can allow family members to know whether they carry the variant causing the disease	2,854	2,106	423	120	66	**5,569**
There is an opportunity to join a patient support group or an online community	2,462	2,159	704	152	92	**5,569**
There is NO opportunity to join a patient support group or an online community	1,253	1,249	1,624	819	624	**5,569**

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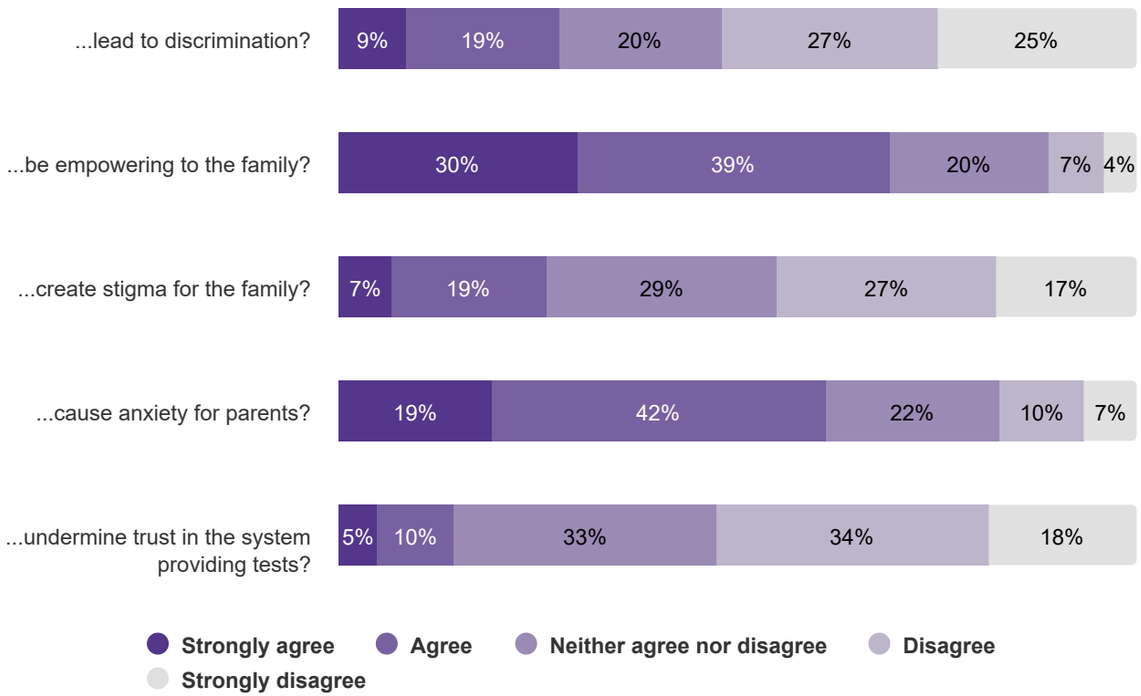
Sample size 5569 responses

In your opinion, could screening for ANY RARE DISEASE at birth...



In your opinion, could screening for ANY RARE DISEASE at birth...

	STRON... AGREE	AGREE	NEITHER AGREE NOR DISAG...	DISAG...	STRON... DISAG...	TOTAL
...lead to discrimination?	482	1,069	1,141	1,505	1,372	**5,569**
...be empowering to the family?	1,682	2,181	1,101	384	221	**5,569**
...create stigma for the family?	388	1,073	1,616	1,522	970	**5,569**
...cause anxiety for parents?	1,083	2,331	1,216	577	362	**5,569**
...undermine trust in the system providing tests?	278	538	1,841	1,893	1,019	**5,569**

Thank you!

For any questions, please contact the Rare Barometer team at
rare.barometer@eurordis.org