



We have an
International
Registry!
Woo Hoo!



Homepage

Home

About

News

Glossary

Explore

My Profile

Coordinators

Approve Users

No Pending Actions



Newsflash

NEW

Genetic Testing

Please upload the patient's genetic test results at the end of the survey. Results can be scanned into a computer and uploaded by using the Attachments tab. Results may also be faxed to the registry at 515-964-1512 or mailed to Dup15q Alliance PO BOX 674 Fayetteville, NY 13066. If you do not have a copy of the results, contact the person's doctor to ask for a copy or contact the registry to ask for help to locate a copy. Results are required to complete your account.



[Click here to register now!](#)

Welcome to the Dup15q Alliance International Registry. The purpose of the registry is to develop a comprehensive database of individuals with Dup15q Syndrome, to better understand the characteristics of the syndrome, determine areas that need further research and develop targeted treatments to improve the lives of those affected by Dup15q Syndrome. The registry has been developed by the generous support of friends and family of the Dup15q Alliance. To learn more about the Dup15q Alliance, please click here, www.dup15q.org

Registration – IRB approved consent process

Understanding Your Participation

For the purpose of this Consent "the affected individual" will refer to the person diagnosed with Dup15q Syndrome. "You" will refer to the person entering the information. This may be the affected individual or a family member or guardian of the affected individual (the person legally responsible for the care and maintenance of the affected individual).

We are asking you to enter personal and other information into the Dup15q International Registry (which we will call The Registry). It is important that we explain what is involved and what will be done with the information you provide. This section contains answers to questions about the information we want, why we want the information, how it might benefit you, and possible risks of giving us this information. After you have reviewed "Understanding Participation" you will be directed to the Patient Consent form. You will be asked a series of questions. If you answer, "yes" to these questions regarding participation and use of the information you are providing you will have "consented." Participation is optional. If you have questions that are not answered in this document, please contact the registry coordinator at: coordinator@dup15qregistry.org.

What is a registry?

A registry is a place to store detailed information about affected individuals with a specific disease or syndrome. In this case, the Dup15q International Registry is for affected individuals with Dup15q Syndrome. Establishing The Registry addresses two critical needs. First, scientists studying Dup15q need accurate, firsthand information to understand how Dup15q affects people. Second, scientists who are ready to start studies, such as those who test new treatments, will need to access The Registry to identify people that may be eligible to participate in studies. If an affected individual looks like a good match for a scientist's research the scientist will contact the Dup15q Alliance. The Dup15q Alliance or the alliance's agent (such as genetic counselor) will then contact the Dup15q family. Scientists will not contact families directly.

What are the benefits of collecting affected individual's information in a registry?

Although there is no direct benefit to any individual participating in the registry, by collecting information on affected individuals in The Registry, scientists can:

1. study why individuals have different symptoms.
2. learn about how certain treatments work and don't work.
3. help medical professionals improve how they treat affected individuals with Dup15q.
4. speed up research in Dup15q by collecting information that scientists can use.
5. let affected individuals (or their family) know when they may be eligible for clinical research studies or clinical trials.

Whose data are you collecting in The Registry?

Individuals who have a diagnosis of Dup15q 11-13, including those who have passed away. The diagnosis must be confirmed by genetic testing.

Consent and Communication Preferences

Registration and Consent

Informed Consent

For the purpose of this document “you” and “your” refers to the registrant, either the individual affected by Dup 15q (affected individual) or the parent, guardian or family member providing the information on behalf of the affected individual (the person legally responsible for the care and maintenance of the affected individual).

1. Your information will be saved in the Registry using a code. The code is used so others don't know who you are. The Registry has processes in place to protect your identity. The Registry may share your coded information with other registries or databases. This information may be used for research or to plan clinical trials. Do you give your permission for your information to be transferred to other registries and databases?

 Yes

2. Your participation in this project is entirely voluntary. Should you change your mind and wish to withdraw your data from The Registry, you will be free to do so without having to provide any explanation. Do you understand this?

 Yes

3. The Registry has been fully explained to me. I understand the “Understanding my participation” and informed consent form. I also know how to access this document in the future if I want to review it. I have had the opportunity to ask questions of The Registry Coordinator. All my questions have been answered to my satisfaction.

 Yes

4. If researchers learn anything interesting about your condition, do you want to be contacted by the Registry with this information?

Yes No



5. The Registry may get information about a clinical trial that you might be eligible for. Do you want to be contacted with this information? *(Please note that even if the coordinators of a clinical trial believe that you might be eligible for the trial, based on the data about you stored in The Registry, it is still possible that later on it will turn out that you do not meet the trial inclusion criteria after all. Please also be*



How do we
Qualify our
Participants?



Account Set-up

Name of Person Creating Account

To begin the registration process, please create a user account below. Once an administrator has approved your account, you will receive an email asking you to log into the system to complete the registration process and provide information about the affected person.

Your First Name: ⓘ ⓘ

Last Name: ⓘ

Email: ⓘ ⓘ

Relation to Patient: ⓘ ⓘ

Username: ⓘ ⓘ

Password: ⓘ ⓘ ⓘ ⓘ

Contact

Address:

City:

State:

Province (if outside USA): ⓘ

Zip/postal Code: ⓘ

Alternate Email:

Country: UNITED STATES ⓘ

Home Phone:

Cellphone:

Second Phone:

ⓘ [Accept Terms and Conditions](#)

Register

ⓘ Required field | ⓘ Information: Point mouse to icon



When a participant is qualified what happens next?

Participant Account

Hi, Coordinator [Logout](#)

[Home](#) [About](#) [News](#) [Glossary](#) [Explore](#) [My Profile](#) [Coordinators](#)

Coordinator Account

Registered By: Vanessa Rangel Miller (Self)

[Update Status](#)

PID: 2789

Clinical Status : Not Reviewed

Genetic Status : No Results Received

If deceased, date of death : -

NOTIFICATIONS

Genetic Test Results Reminder

Please submit a copy of your genetic test results. If you have a copy of your results in an electronic format, you may attach them to your profile by selecting the Add Attachment link below.

[Profile](#) [Physicians](#) [Contact Info](#) [Family](#) [Attachments](#) [Testing](#) [Contacts](#) [Newsletters](#) [Notes](#)

[Clinical Questionnaire](#)

 Completed: 2011-08-18

Add Attachments

Clinical Status : Not Reviewed

Genetic Status : No Results Received

If deceased, date of death : -

NOTIFICATIONS

Genetic Test Results Reminder

Please submit a copy of your genetic test results. If you have a copy of your results in an electronic format, you may attach them to your profile by selecting the Add Attachment link below.

Profile Physicians Contact Info Family **Attachments** Testing Contacts Newsletters Notes

A copy of the patient's genetic test result is required to complete your account. You can submit other records if you choose including reports from the following assessments: sleep study, EEG, MRI, muscle biopsy, etc. The Registry Coordinator may need to contact you to clarify the genetic test results. If you need help to locate the genetic test results, please [contact the Registry Coordinator for assistance](#). Results may also be faxed to the registry at 515-964-1512 or mailed to Dup15q Alliance PO BOX 674 Fayetteville, NY 13066.

To attach electronic attachments, including scanned images and electronic files, please select the type of attachment below, choose a file from your computer and upload. File types must be in the following formats: Acrobat (pdf), Microsoft Word (doc), Microsoft Excel (xls), or JPEG Image (jpg).

Type: **Cardiac/Heart Test Results (ECHO/EKG)** Choose a file to upload

- Cardiac/Heart Test Results (ECHO/EKG)**
- Developmental Report
- Genetic Test Results
- Muscle Biopsy
- Neurological (brain) EEG
- Neurology (brain) MRI
- Sleep Study
- Other

Type	Description	Date Created
------	-------------	--------------



**A little bit about the survey content
and layout...**

Survey Topic Areas

Anesthesia	3
Behavior	8
Cardiac	5
Cognitive Development	7
Deceased	11
Development	13
Diagnosis	8
Ears/Nose/Throat	7
Gastrointestinal	7
General Health	5
Immunology/Allergy	4
Neurological	6
Neurological - Seizures	16
Orthopedic	4
Pregnancy/Neonatal	11
Puberty	6
Pulmonary/Sleep	9
Renal/Metabolic	7
Survey Comments	3
Vision	3

Correct Responses



Coordinator Account

Clinical Questionnaire

[Re-take the survey](#)



Completed: 2011-08-18

Click on a section heading below to view and update your responses and view responses from all users.

Click to update a single response, click to see how your responses compare to other members of the community.

Diagnosis

What is the affected person's chromosomal diagnosis?	Unsure
How old was the affected person at the time of Dup15q Syndrome diagnosis?	No Response
Has the affected individual ever been formally tested for an autism spectrum disorder?	Yes
Has the affected person been diagnosed with any of the following?	None
At what age was the affected person diagnosed with the following?	No Response
Has the affected person ever had seizures?	No
Does the affected person have a blood sample stored at Coriell?	No Response
Does the affected person have siblings? (check all that apply)	No Response

Complete Survey

Coordinator Account

You do not have to complete the survey at one time. You may log out at any time and your answers will be saved. For more detailed instruction, [click here](#).

Pages Clicked

40%

Cognitive Development

What non-verbal communication skills has the affected person ever used on a regular basis? (check all that apply)

- Gesturing
- Pointing
- Sign language
- Picture Exchange Communication System (PECS)
- Icons or symbols for communication (real photos or icons to represent words, actions or activities)
- Voice Output Communication Device
- None
- Unsure
- Other, please explain :

How many words are in the affected person's vocabulary?

- 0 words
- 1 - 10 words
- 11 - 20 words
- 21 - 30 words
- More than 30 words
- Unsure

Please indicate the affected person's level of verbal communication.

Participants Explore Data

Explore Responses

The following is a summary of all profile responses. Click on a topic area from the listing below to view detailed responses of all registry participants.

[Diagnosis](#)

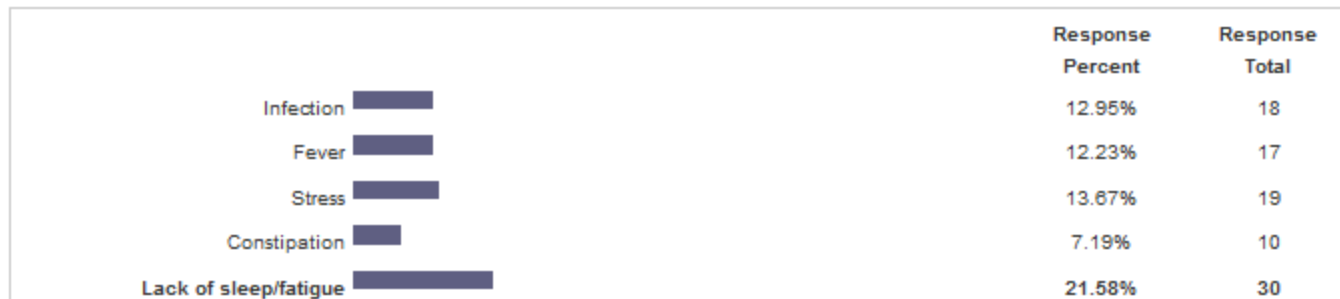
[Neurological - Seizures](#)

click the title again to close

What time of day do the seizures most often occur?



Which of the following factors make the seizures worse? (check all that apply)



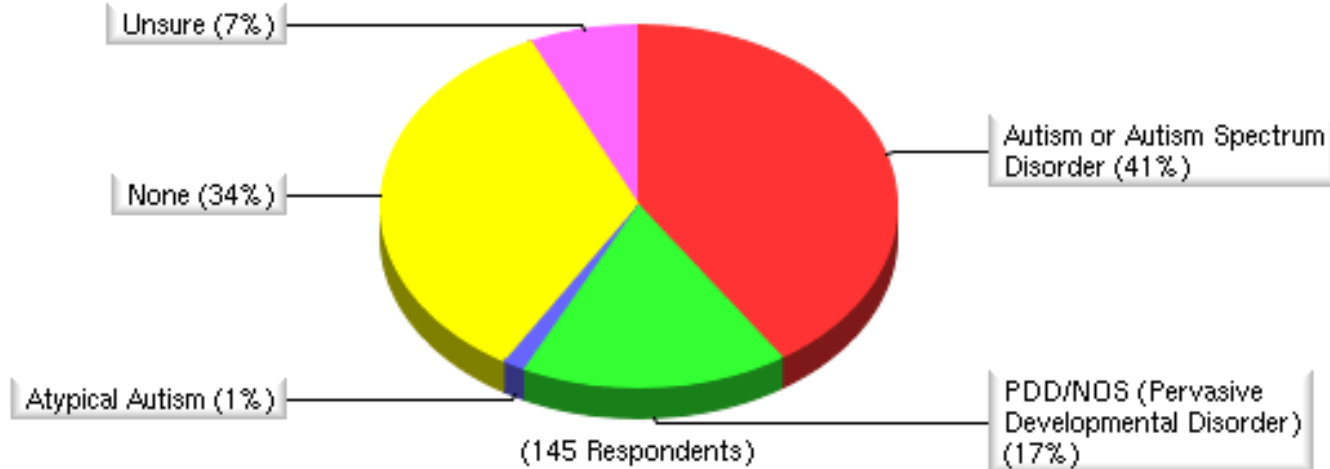


Some interesting things we are seeing...

What is the affected person's
chromosomal diagnosis?

Autism Diagnosis

Has the affected person been diagnosed with any of the following?



Diagnosed with ADD/ADHD

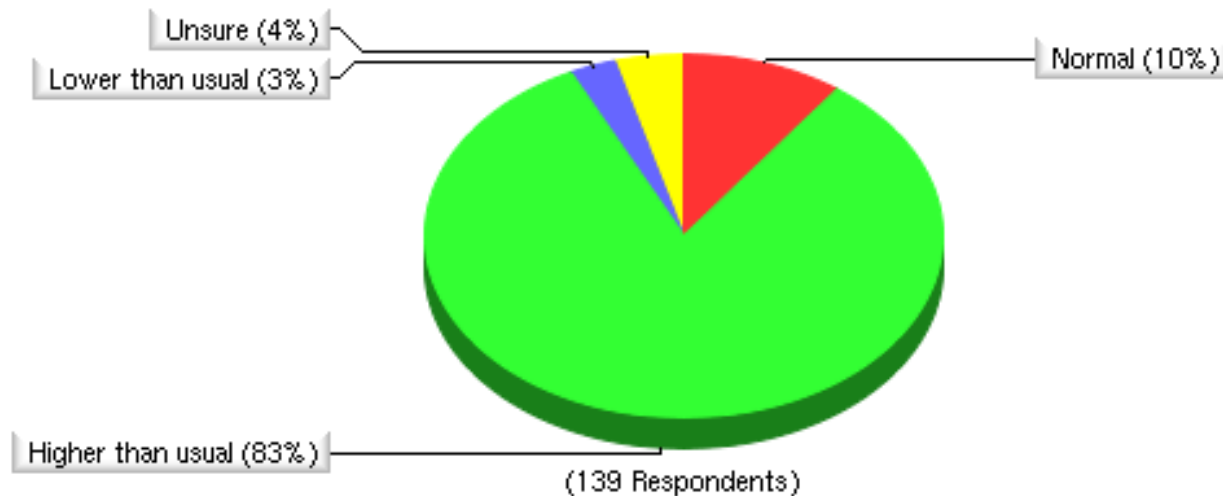
Has the affected person ever been diagnosed with ADD/ADHD?



Supportive therapies

Pain tolerance level

What is the affected person's pain tolerance level?



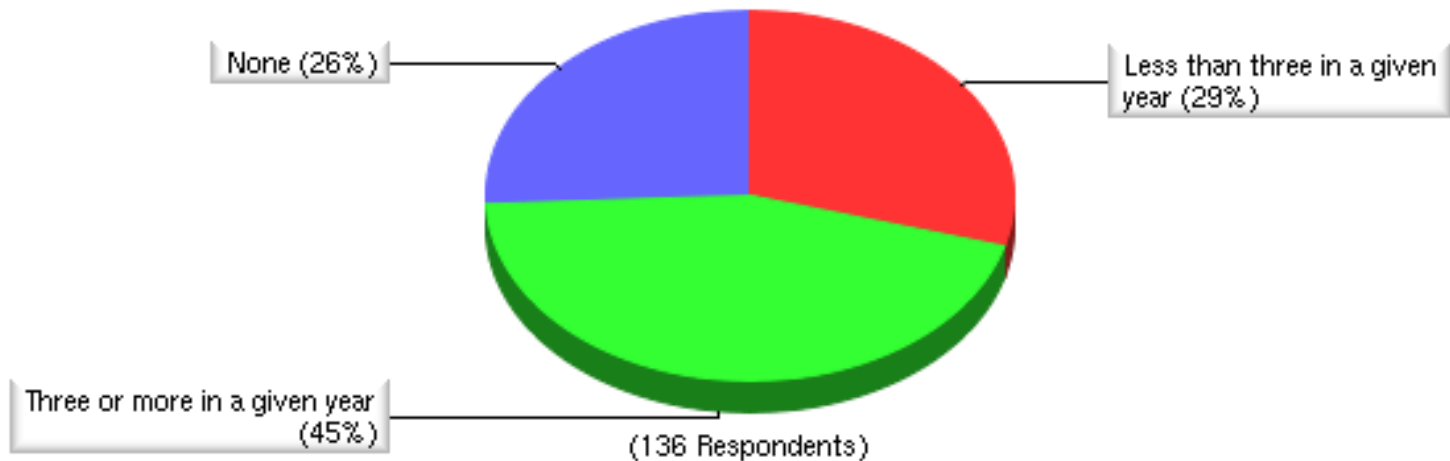
Seizure – Loss of Skill

Have the seizures caused a loss of skill or ability? (check all that apply)

	Response Percent	Response Total
Loss of motor skill(s) (gross or fine motor)	23.81%	25
Loss of complex skill(s) (e.g. toilet training, reading)	12.38%	13
Loss of speech (e.g. individual words, or sentences)	13.33%	14
None	24.76%	26
Unsure	15.24%	16
Other, please explain	10.48%	11
Total Respondents :		78

Ear infections

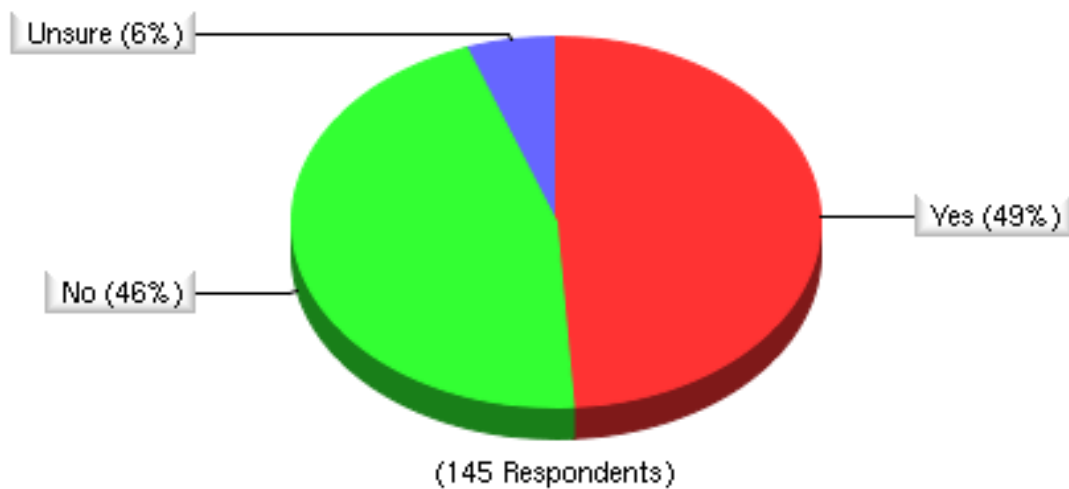
Has the affected person had frequent ear infections, currently or in the past?



Age at Diagnosis

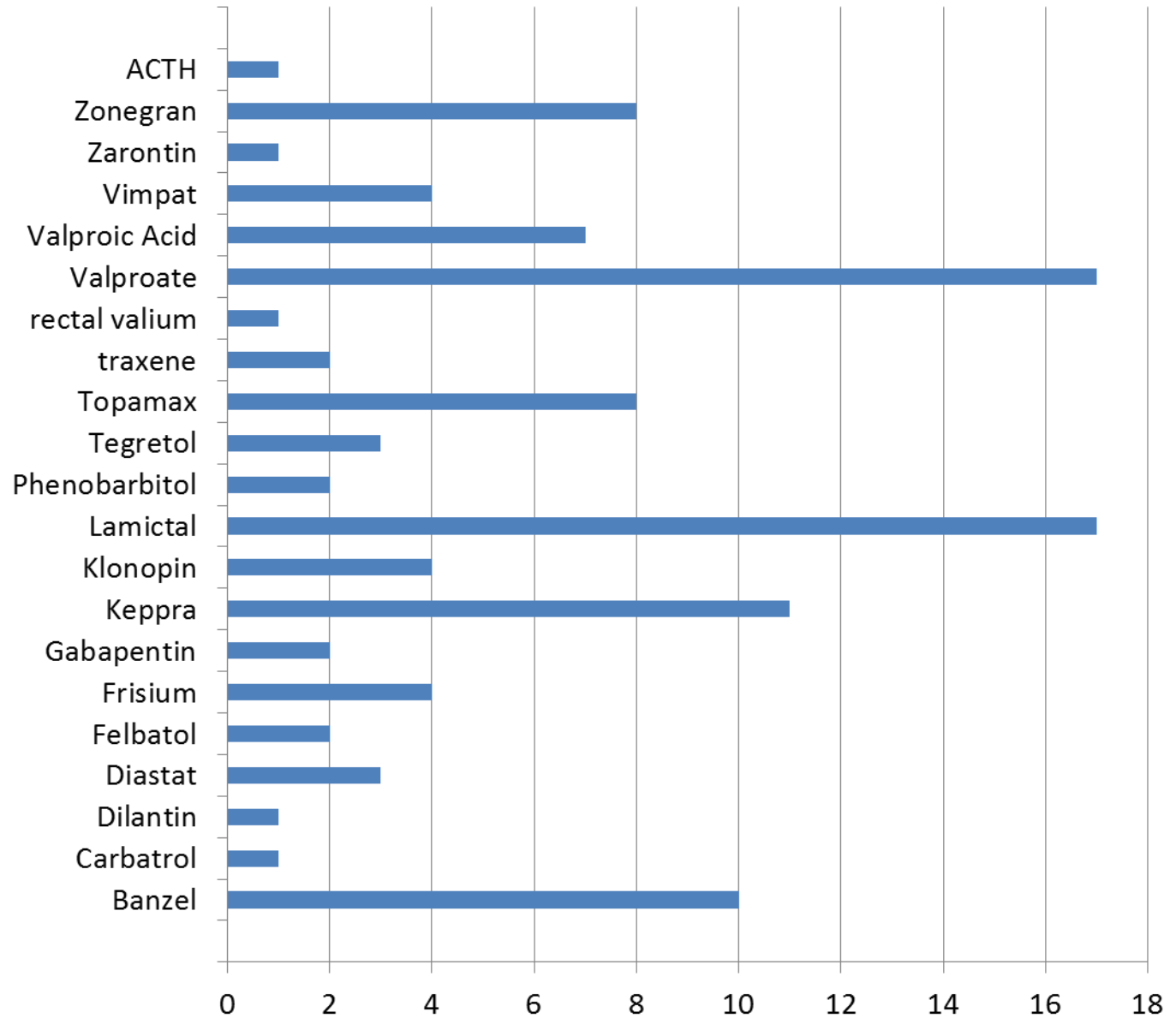
Seizures

Has the affected person ever had seizures?

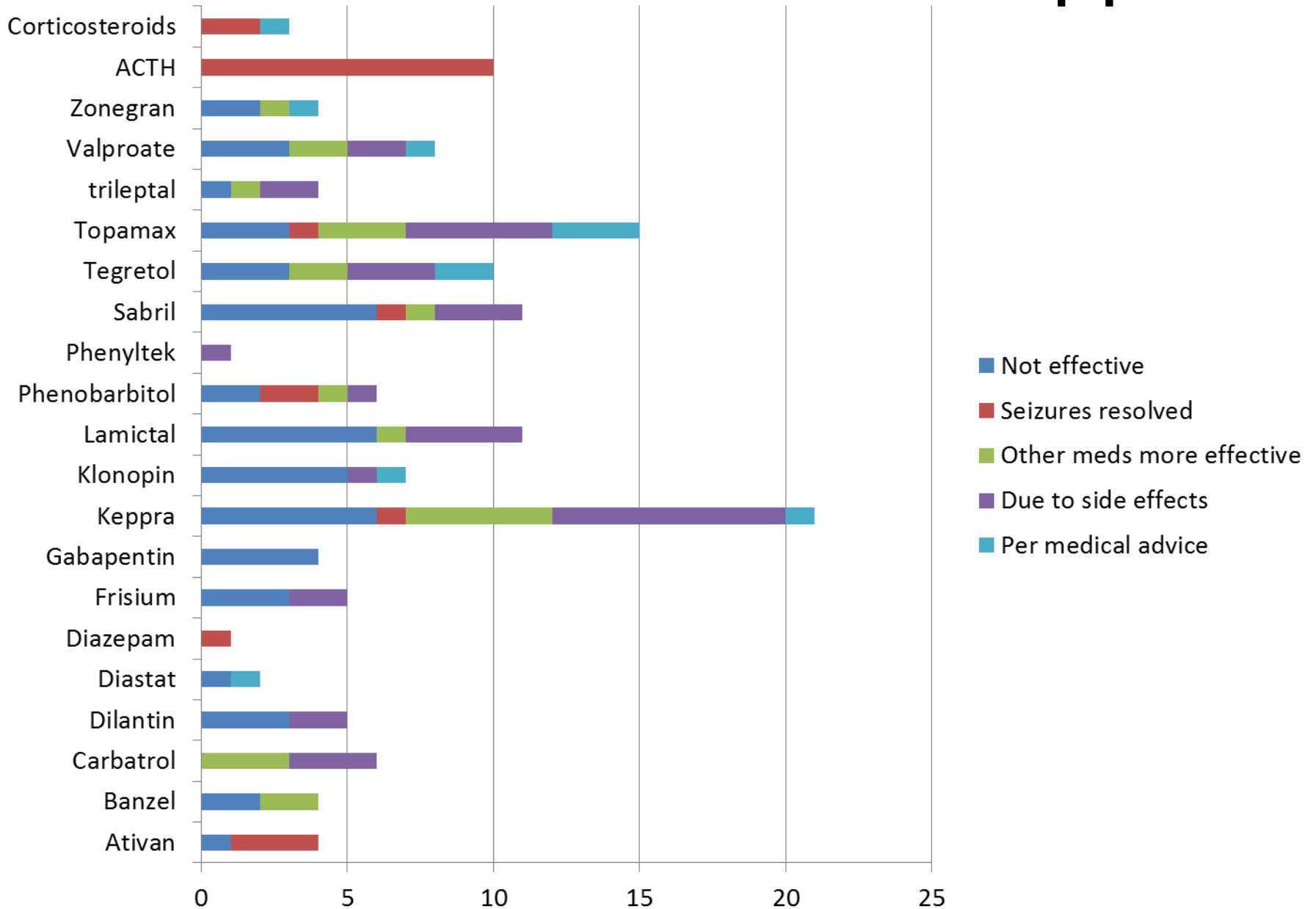


Seizure medications – Current usage

Currently taking

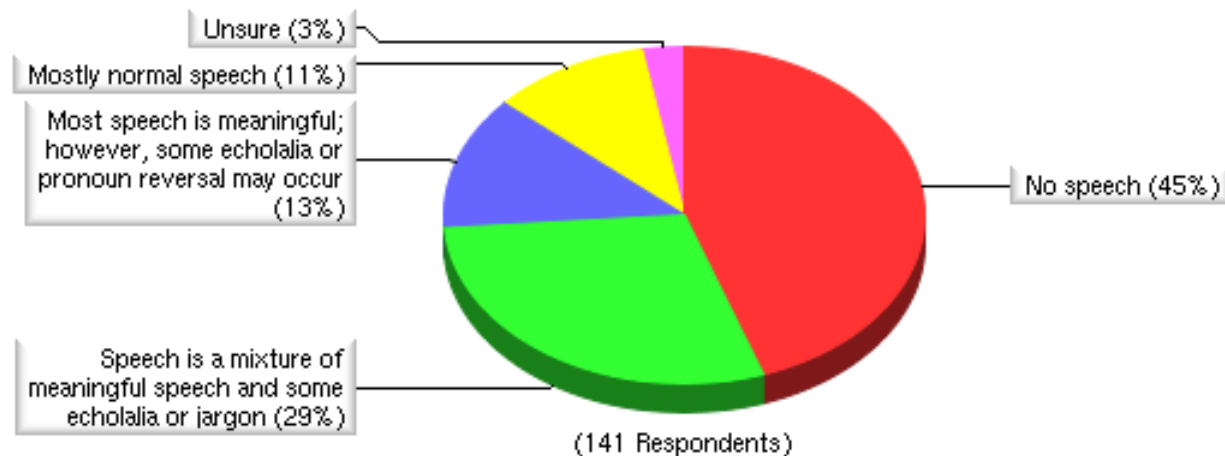


Seizure medications - Stopped



Level of verbal communication

Please indicate the affected person's level of verbal communication.



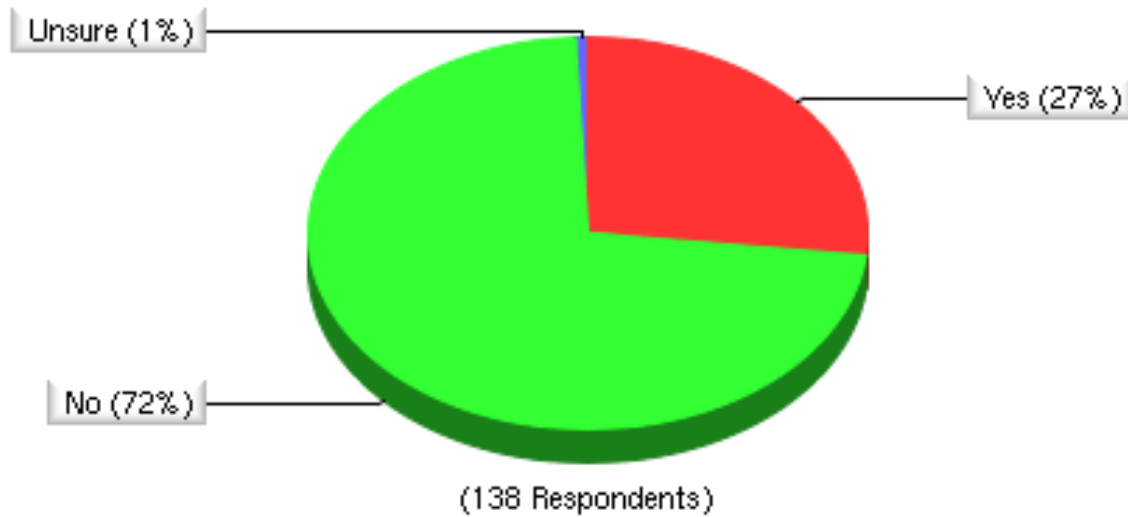
How many words are in the affected person's vocabulary? NZ

How many words are in the affected person's vocabulary?



Sleep study

Has a sleep study been done?



Sleep study results

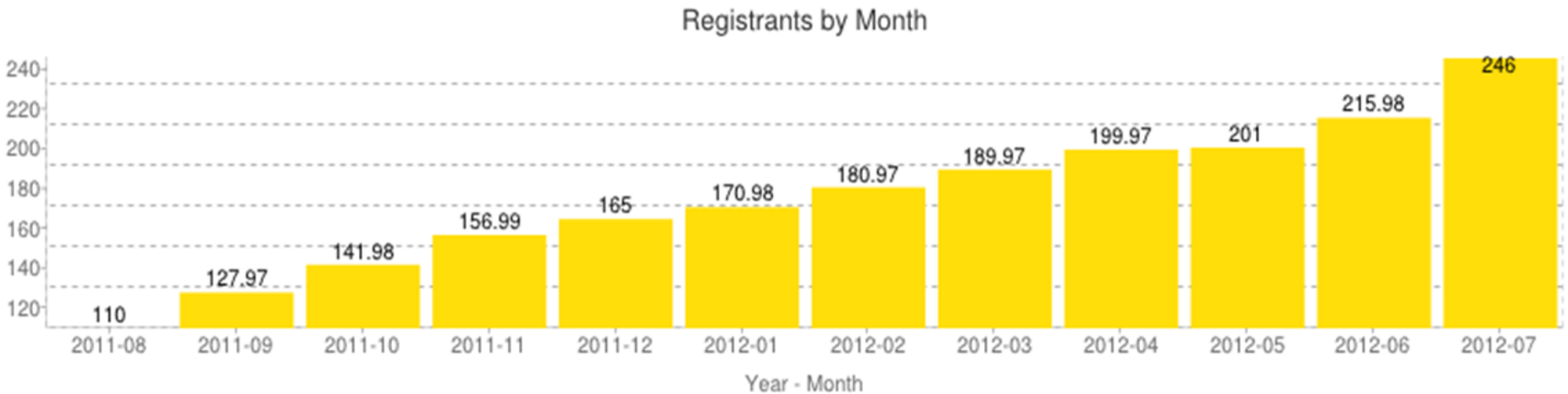
If the affected person had a sleep study, what were the results?

	Response Percent	Response Total
Not applicable - has not had a sleep study	60.17%	71
Normal	8.47%	10
Mild sleep apnea	11.02%	13
Severe sleep apnea	0.00%	0
Central apnea	1.69%	2
Lack of REM	5.93%	7
Unsure	2.54%	3
Other, please explain	10.17%	12
Total Respondents :		112

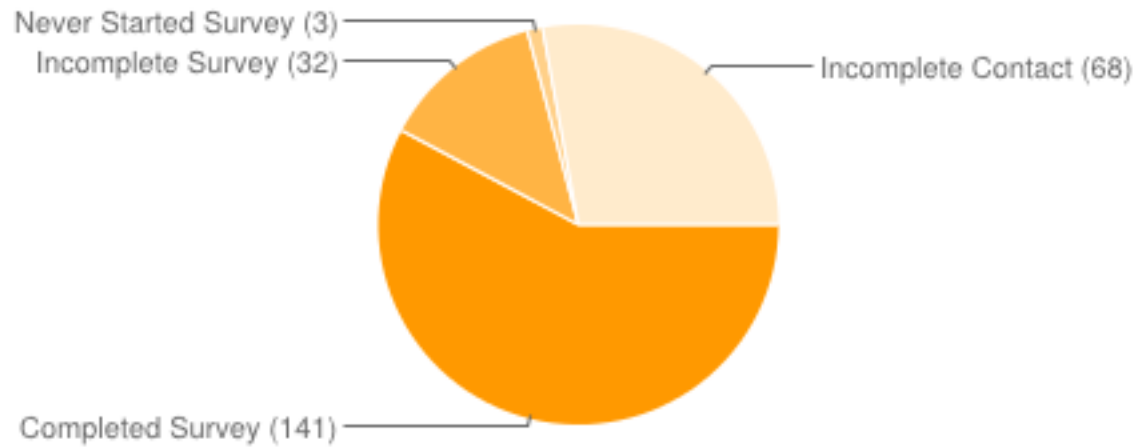


Some of our demographics
include...

Registrants by Month

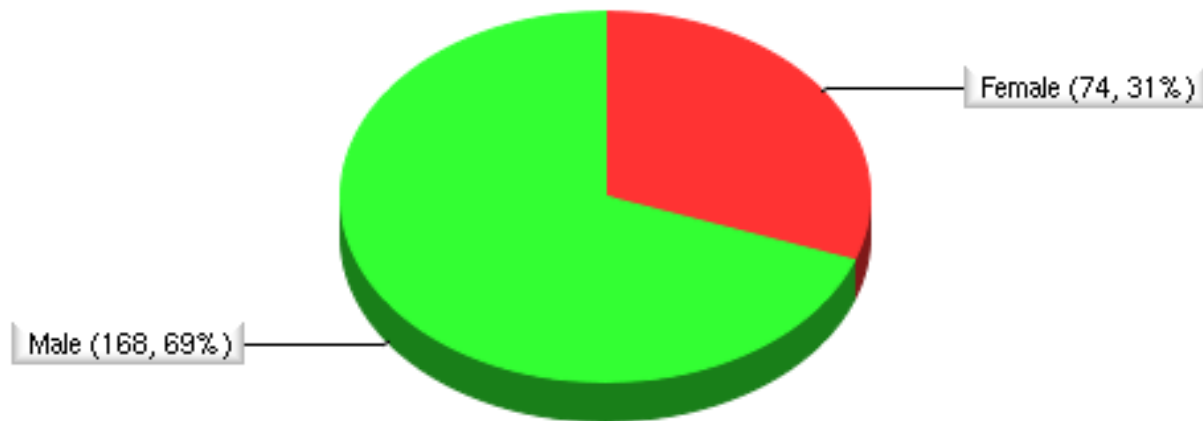


Profile Survey Status



Gender

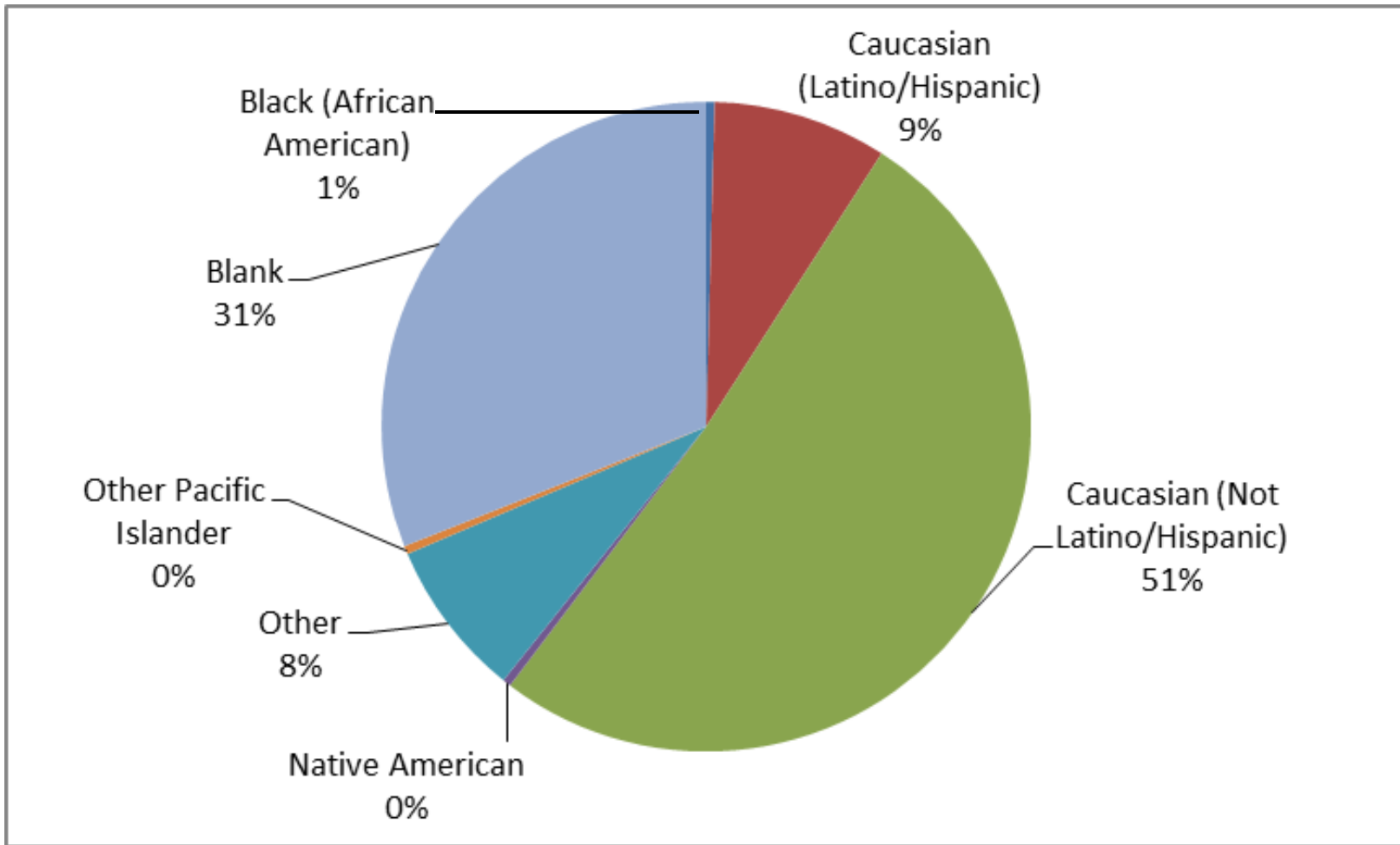
Gender



Country of Residence

ARGENTINA	2
AUSTRALIA	16
AUSTRIA	2
BELGIUM	1
CANADA	16
FRANCE	1
GERMANY	1
IRELAND	3
ITALY	5
MEXICO	1
NORWAY	1
PERU	1
SCOTLAND	1
SPAIN	5
UNITED KINGDOM	24
UNITED STATES	162
Grand Total	242

Ancestral Background



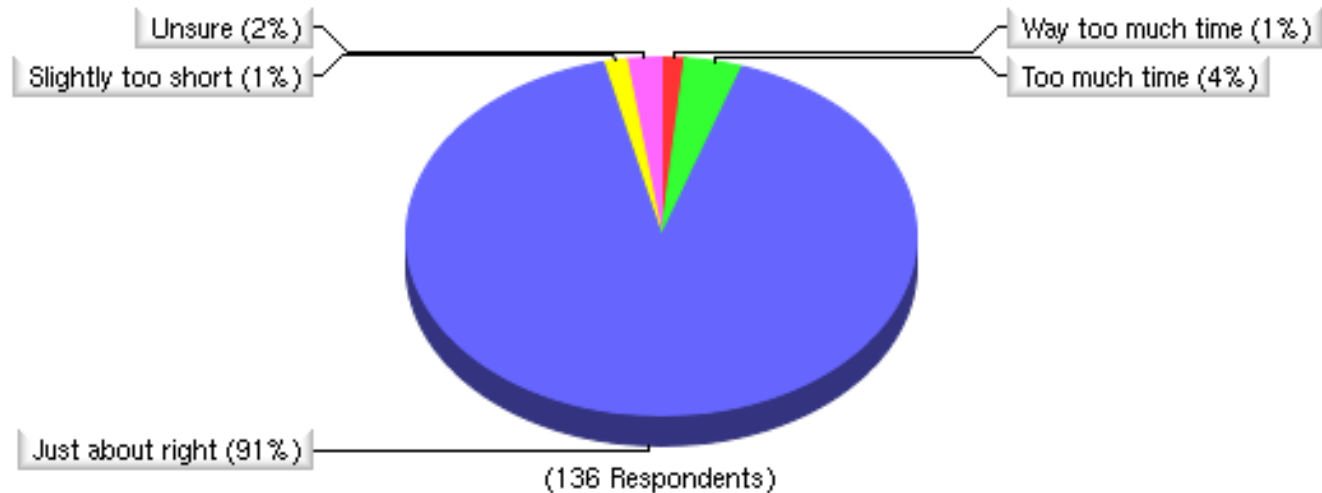
Current Ages (n=173)



Feedback about the survey

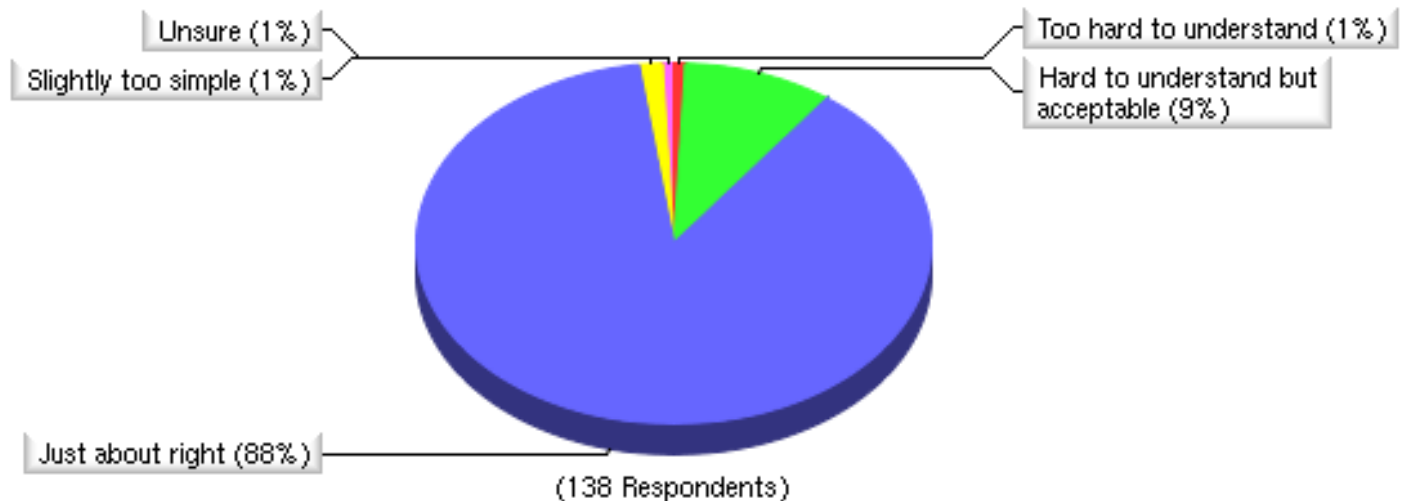
How long did the survey take to complete?

How long did the survey take to complete?



How easy was the survey to understand?

How easy was the survey to understand?



DUP15q
ALLIANCE

INTERNATIONAL REGISTRY

Whew! We
are all
done. Let's
discuss!

