2014: an Atypical HUS Global Poll Collaboration

SURVEY QUESTIONS

2014 aHUS Global Poll: Survey Questions

In 2014, the aHUS Alliance collaborated with the EURORDIS sponsored RareConnect team to conduct a survey of adult atypical HUS patients and family caregivers of pediatric aHUS patients. (N= 217 participants from 17 nations)

While the RareConnect platform of rare disease communities was retired in 2023, some poll assets were previously archived and remain available. Unfortunately, the actual 20214 tool is not available but here are the questions involved in that global poll.

An archived whitepaper written regarding the 2014 poll data and analysis is available here: https://bit.ly/2014aHUSpollWhitepaper

A EURODIS PROJECT

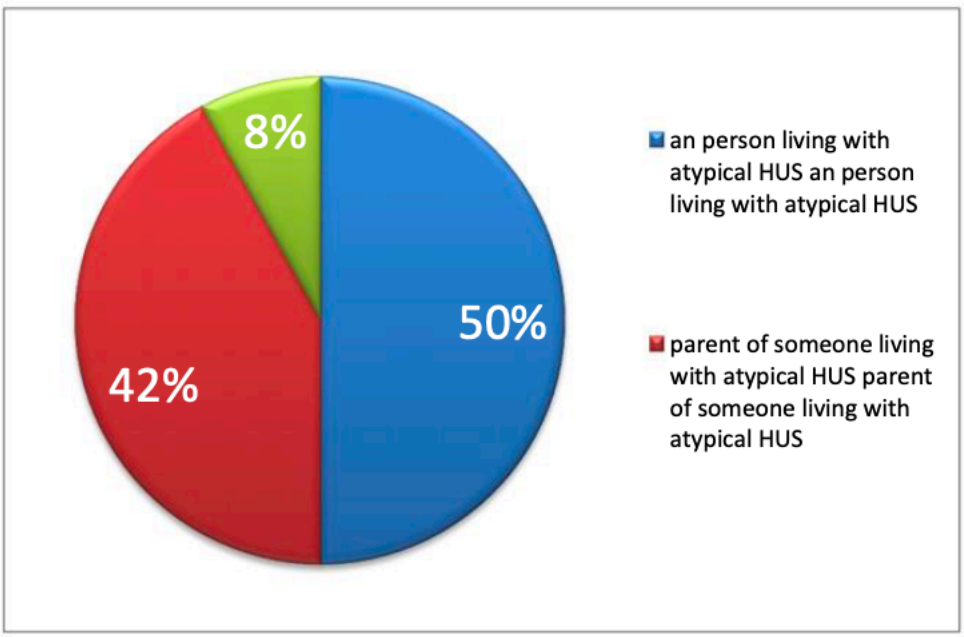
aHUS Survey Results

Description:
The purpose of this poll is to allow members of the atypical HUS community on RareConnect.org to see how we have lived through the process of obtaining a diagnosis and some of the consequences of the disease on our lives.

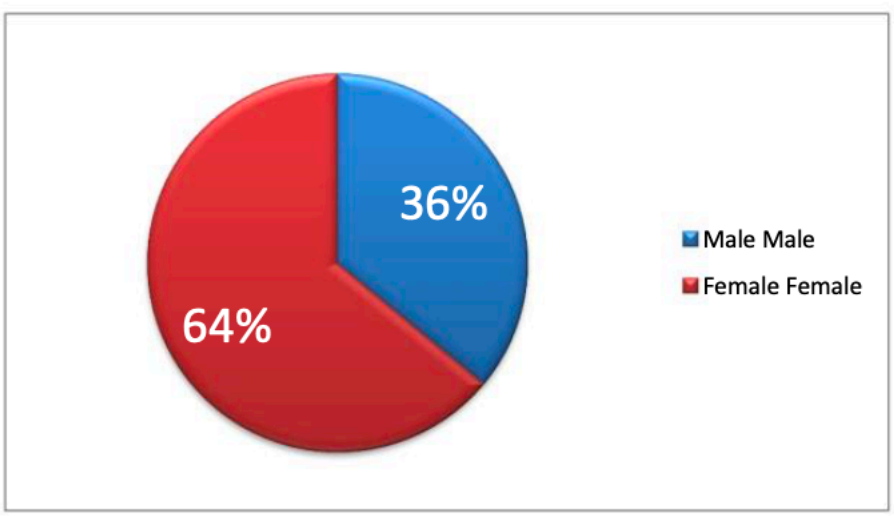
Total number of responses: 214

Survey open from 2/17/2014-3/07/2014

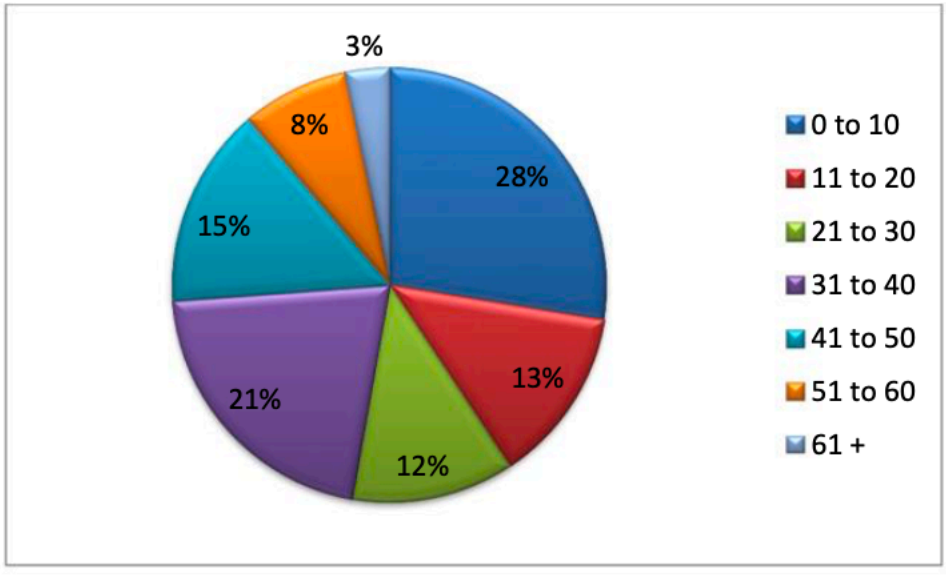
1. The person filling in this form is:



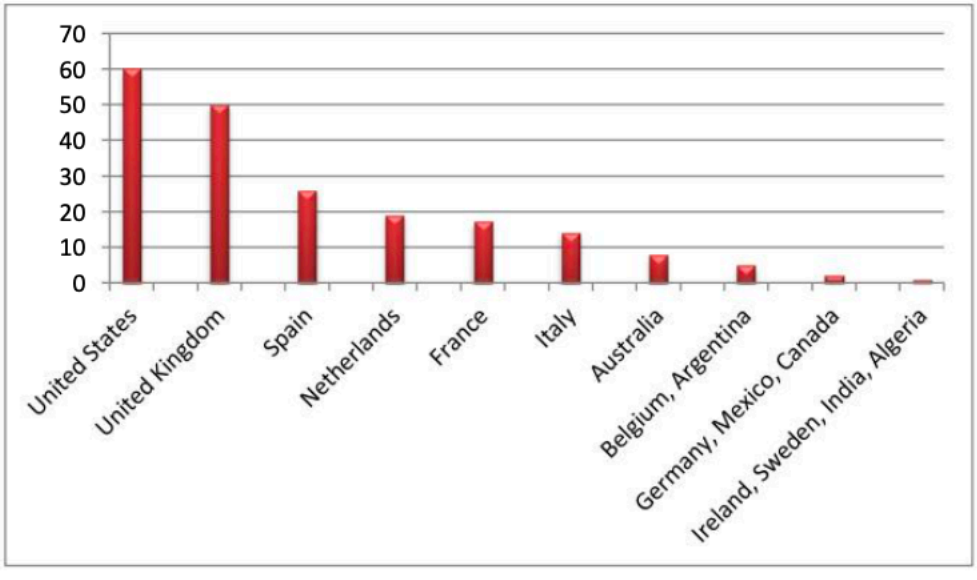
2. Gender



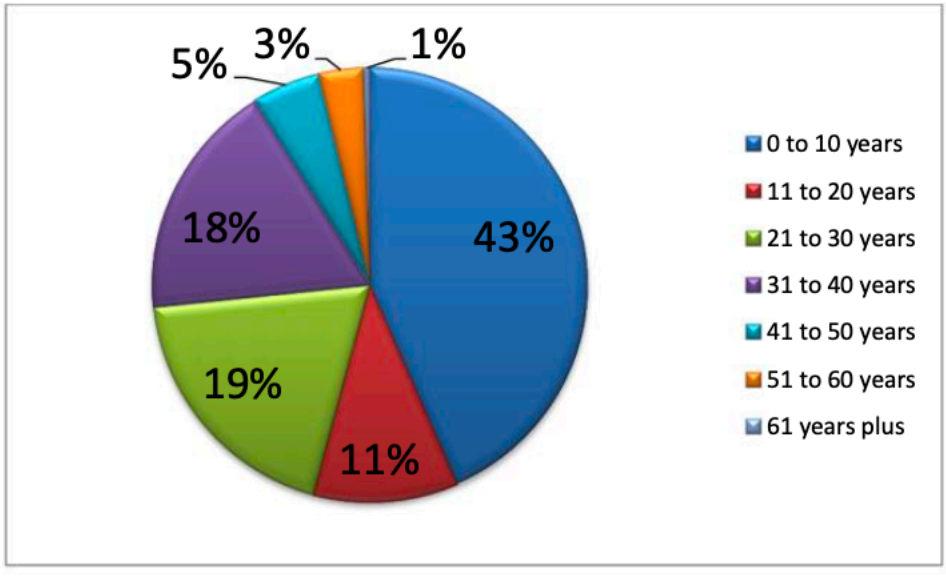
3. Age



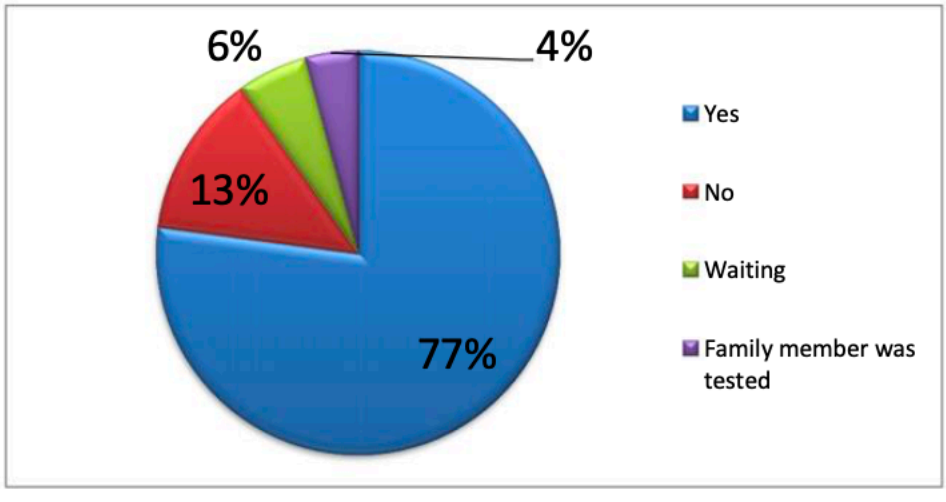
4. Country of residence



5. Age when became ill with aHUS



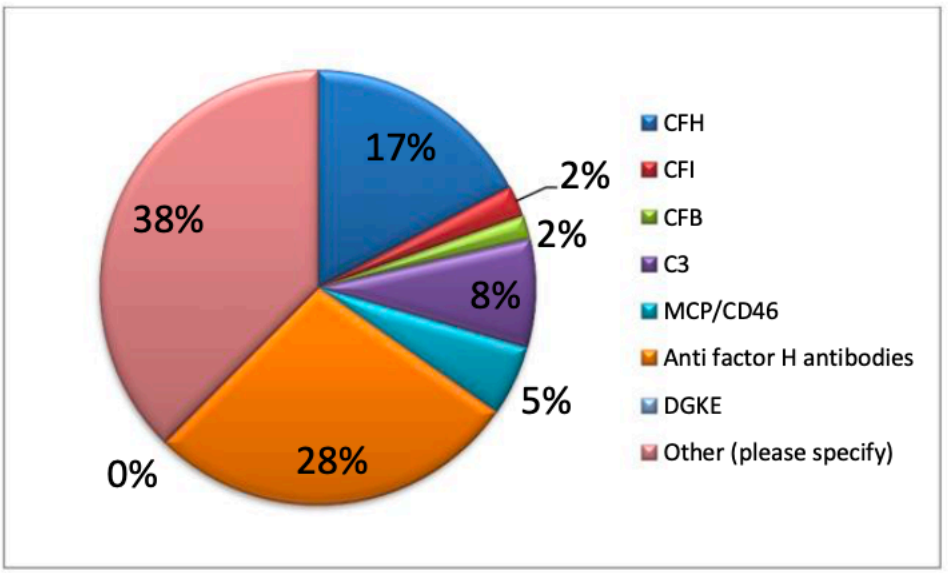
6. Have you undergone genetic screening?



Comments:

- Don't know
- In process of doing it now
- Both my son and i have AHUS
- Negative
- All family members were tested and shown to have a factor H deficiency
- Factor H, I, and MCP
- CFH

7. If yes, what is the predisposing genetic cause of your atypical HUS?

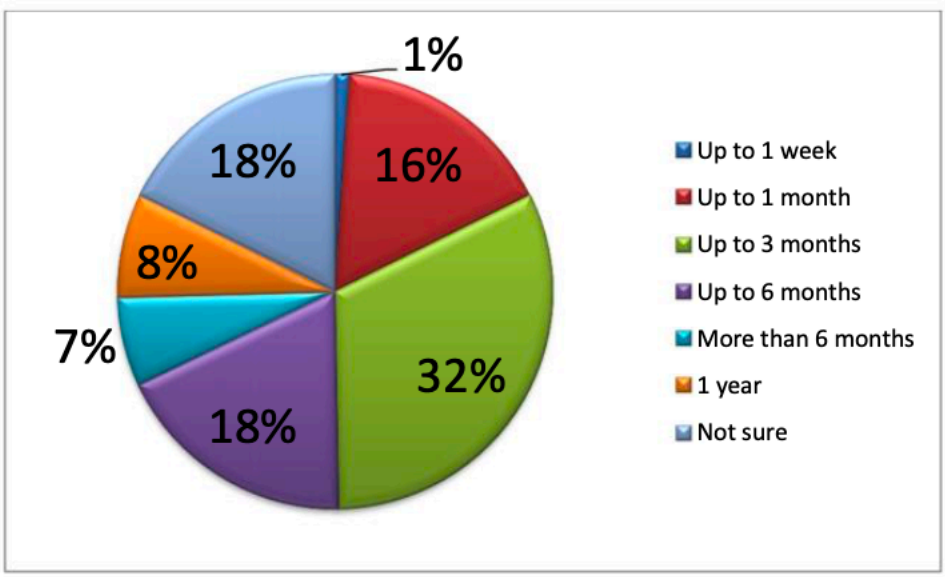


* No response 41

Comments for other:
- Not known
- No genetic mutation identified to date
- H ,g I,
- no conclusion
- no confirmation of a mutated factor
- No genetic cause
- Still unknown
- CHF/CFHR1 fusion
- none found
- Factor I
- not known
- Can't remember the exact it Factor H mutations-heterozigoziz
- not known
- Normal Factor H, Factor I, and MCP. No further testing has been done.
- Unknown
- Don't remember
- N/a
- don't have the results yet
- Not identified
- All genetic testing negative
- CFHR3 deletion and CFHR1 deletion resulting in CFH/CFH 1 fusion protein
- cfhr 1 and 3 deletions. Investigating on a fused mutation.
- Unknown
- I don't have this info on me, university of Iowa has the testing results, can provide if necessary
- Don't know
- unknown
- inconclusive
- Unknown
- All of her genetic testing came back normal
- unknown
- See above
- Unknown still
- MTHFR

- Unknown
- unknown
- CFH/CFHR1 Hybrid
- thrombomodulin and two CFI
- both CFI and C3 + CFH deficit
- unknown
- Resultado normal no hay mutaciones
- No lo se exactamente
- desconocida
- no lo se
- aun por descubrir. mi caso no es genético.
- Ambientales
- r1-r3
- nessuna origine certa
- Attualmente solo il C3 e il C4 risultano bassi, sono in corso ulteriori accertamenti
- Complemento del Fattore H (penso!)
- CFI + Con ne sais pas
- inconnu a ce jour
- connais pas
- mutation du SCR20 du facteur H
- 3 + DEFICIT CFH
- alles uitgesloten
- Is mij niet bekend.
- Hebben ze me nooit verteld
- Weet ik niet
- Ze hadden geen idee.
- Nog onbekend
- onbekend
- niets gevonden

8. If you had genetic screening, how long did it take to receive your results?



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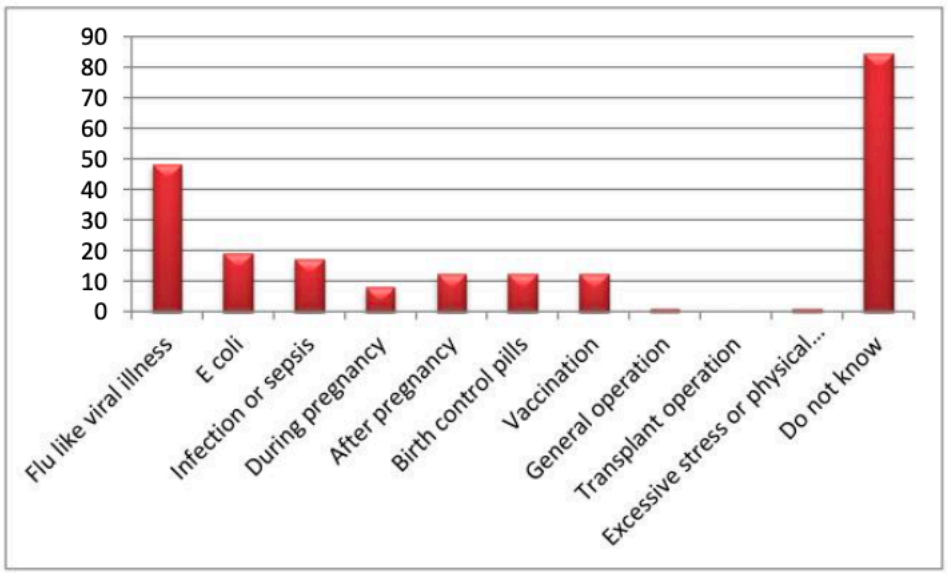
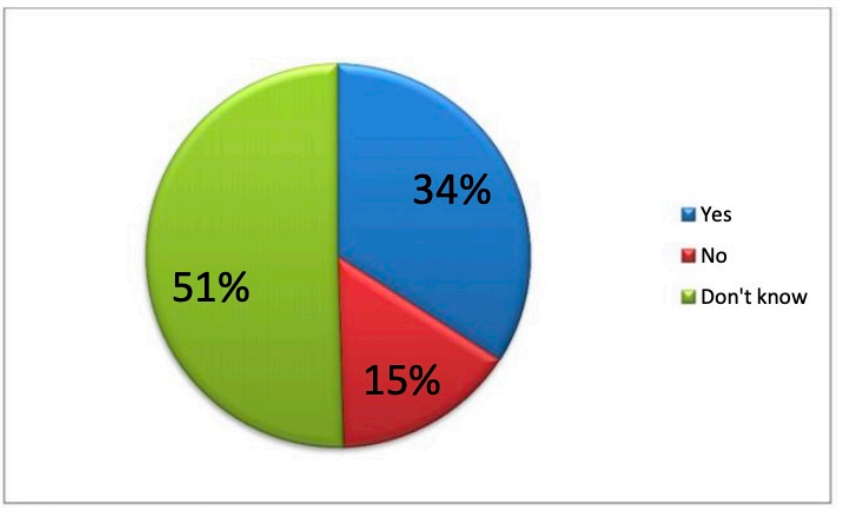
9. What do you assume to be the likely trigger of the first manifestation of atypical HUS?



Other

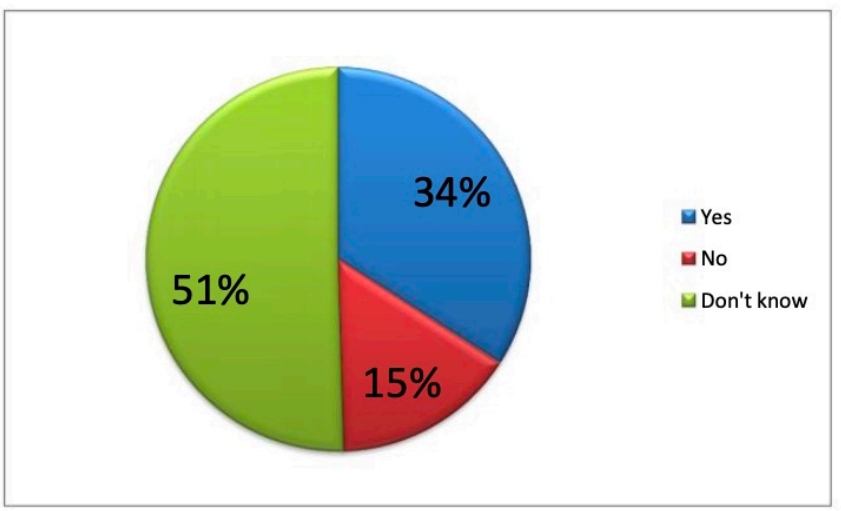
- Shigella infection
- chicken pox
- double cfh gene from father and mother
- Positive for rotavirus (negative for E coli)
- Virus causing vomiting
- HBP. Cat scan with contrast dye
- First pregnancy, vaccination at time of her delivery and birth control pills
- Sickness bug
- pancreatitis
- Gamma Globulin shot after exposure to Measles
- H1N1 vaccination became ill 3 weeks after swine flu vac
- stretch of viral illnesses - ear infections - but not first
- Gastro
- he had a gastro type virus 2.5 months before he was diagnosed
- Citomegalovirus
- vomitos y anemia
- anginas
- Asintomática
- Consumo de cocaina
- sospetta allergia o intolleranza ai frutti di mare
- insufficienza renale
- Ero in terapia con pillola anticoncezionale
- gastro-entérite
- suspicion e coli
- produits toxiques (peintures)
- Infection ORL
- Ernstig bloedverlies door vleesboom daardoor verzwakt en hormonen geslikt.
- Longontsteking of griep of zwangerschap
- Gebruik contrastvloeistof
- samen met flinke verkoudheid
- nierstenen (2 weken vooraf)

10. Were you initially diagnosed with TTP or HUS?

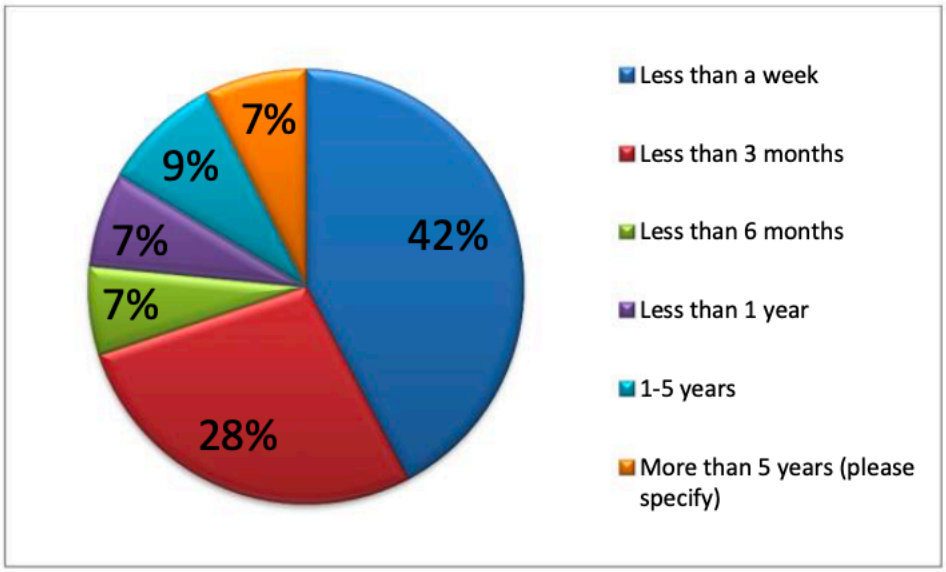


* No response 2

11. Did you receive the Adamts 13 or Shigatoxin test?



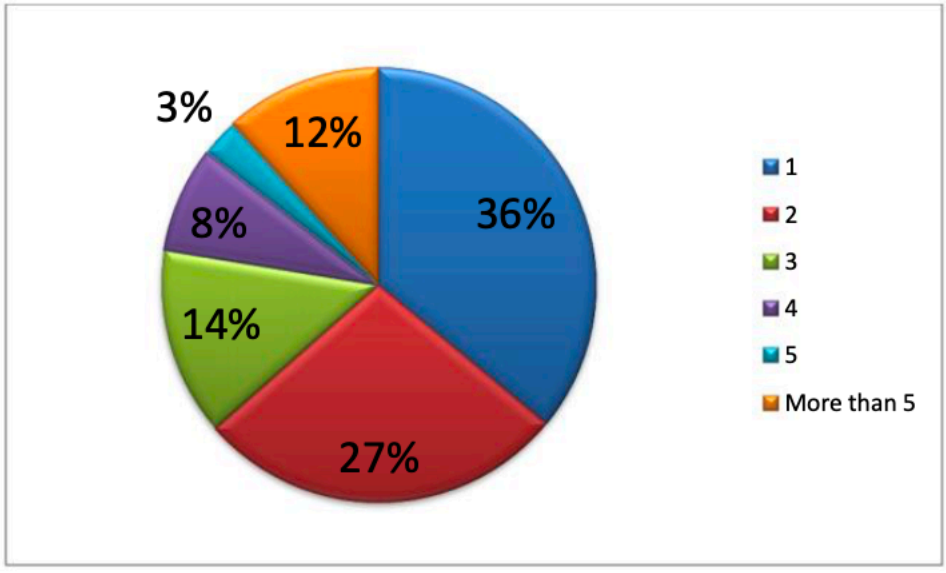
12. Diagnosing atypical HUS is complex. How long did it take before you were correctly diagnosed as having atypical HUS?



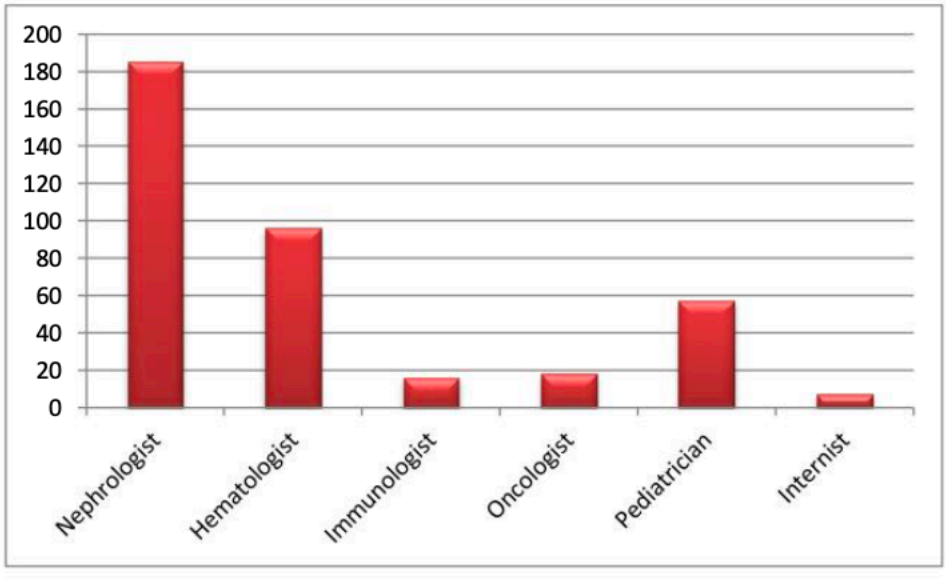
Comments (for more than 5 years):

- Following second episode of aHUS 14 yrs after first onset.
- 8 years
- still cant confirm 100%
- Lack of understanding re diagnosis among healthcare professionals
- Original kidney failure in 94 diagnosed ttp 98
- It took more than 25 years to take the genetic tests, but they told us before they tought it would be - Ahus
- 2 weeks
- 14 years
- 9 years till I was told its what I have
- Still waiting for answers.
- 7 a los 17 años del primer episodio
- Cuando tuve el 2° episodio, casi 8 años después del 1°
- Dal 2004 sono sottoposta a studi presso l'istituto "Mario Negri" di Ranica
- Heb het al jaren, 1 zus overleden, 1 zus in predialyse
- Na de behandeling werd dit niet meer relevant gevonden
- Dat het HUS was wist de arts meteen. Daarnaast had ik ook een andere ernstige ziekte die ook behandeld moest worden. Pas na jaren was ik er aan toe om genetisch onderzoek te laten doen omdat ik toen in aanmerking wilde komen voor transplantatie.

13. How many doctors did you visit before an accurate diagnosis was made?



14. What specialist(s) were involved in your diagnosis at the initial onset? (check all that apply)



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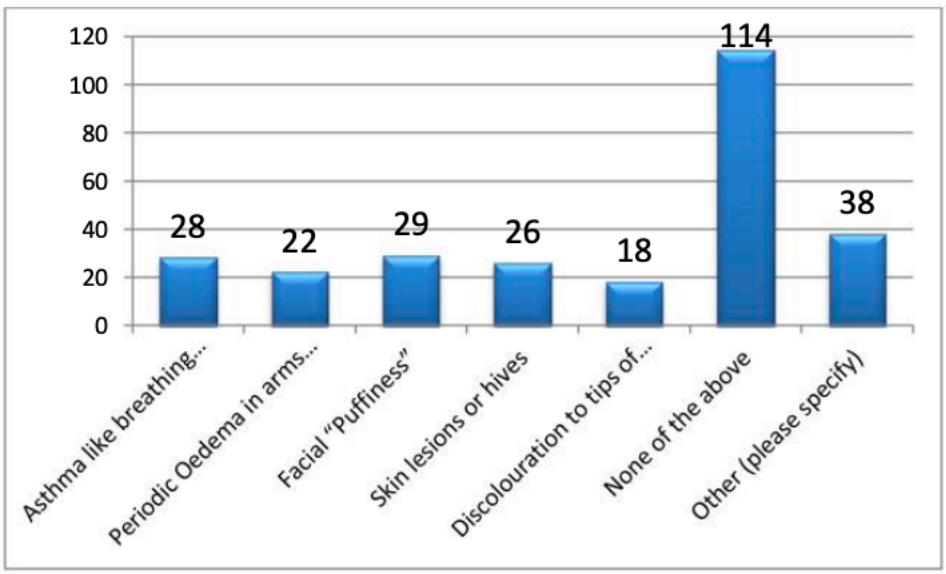
Comments

- im not sure of their titles
- neurologist, cardiologist, physiotherapist
- Geneticists,
- ER doctors
- obstetrician
- Don't remember
- neuology

- It took 6 hours at a children's hospital with an entire team including a liver doctor working together - who came up with the diagnosis
- Gastrologist
- Urologist
- ER Dr. at Buffalo Childrens Hospital
- I do not know I was not told
- GI, Allergist, then once in hospital added nephrologist, infectious disease, neurologist, and hemotologist
- Neuology
- Intensivista
- Cardiologo
- Infectólogo
- Médecin traitant
- Neurologue
- Zou de anderen niet meer weten.
- Én een Professor uit Nijmegen

15. At any time in the years preceding the manifestation of atypical HUS (including childhood) did you experience any unexplained symptoms, such as... (check all that apply)

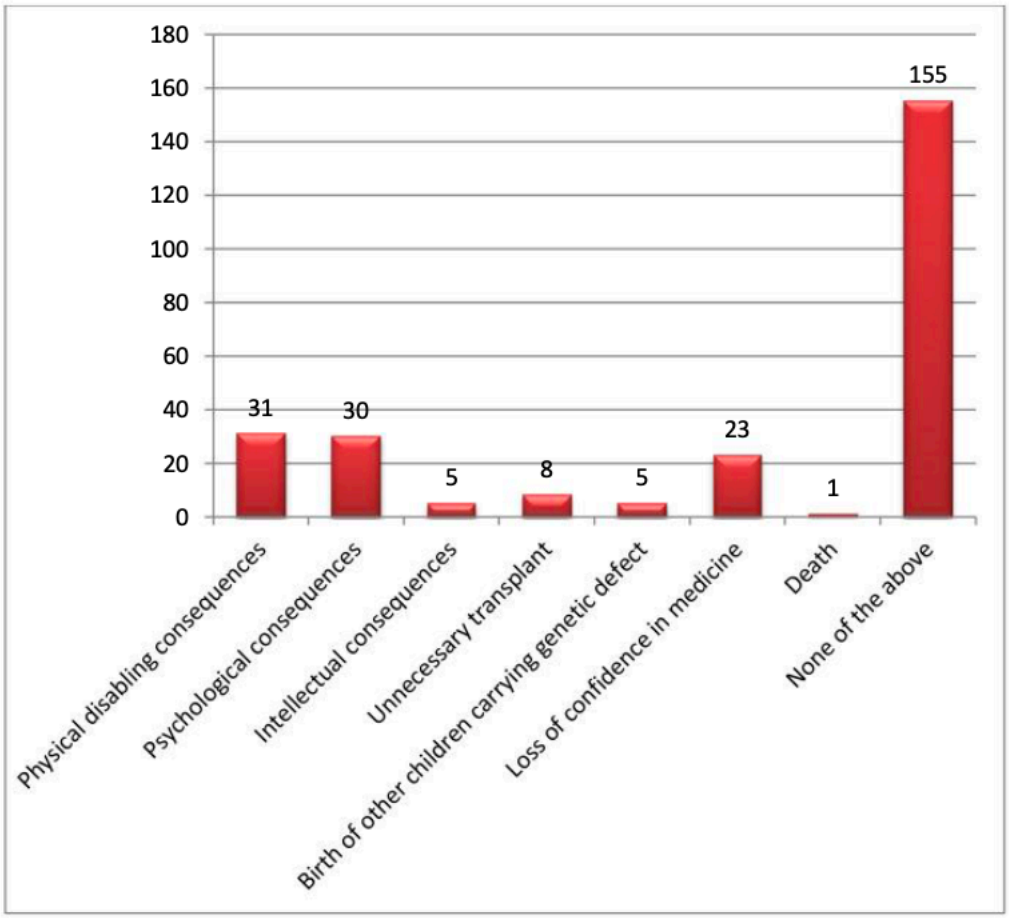


* No response 5

Comments (please specify for Other):
- i have asthma since childhood, cold and tired
- Born hypoglycaemic, polycythaemic, and thrombosytopenic. Also, some neuro symptoms from birth.
- Very often sick
- I am diagnosed asthmatic and had severe eczema
- Jaundice of skin and yellowing around the eyes
- vomitting (including projectile), stopped breathing in cot twice (monitor alarm went off), born hypoglycaemic, polycythaemic and thrombocytopaenic
- dark urine, jaundice, bruises, lethargy
- lower back pain, regular sickness
- frequent bronchitis with swollen outer ears (early childhood)
- Violent Headaches
- quinceys
- constant sickness for no reason and filling ill all the time
- Brusing

- migraines
- a lot of bruises on my legs
- Anemia
- erythema annulare
- Back pains
- extreme lethargy and extreme refusal to eat
- Back pains
- lots of stomach pain and joint pain
- Always being diagnosed with hand food and mouth disease
- bad headaches and sporadic temperature
- hospitalization for low platelets following roseola - resolved with no interention
- family member has aHUS, so we did genetic screening on other son. We were aware in advance to be watchful.
- Fainting
- Dolor en articulaciones inferiores, sangrado nasal
- siempre tuve plaquetas bajas
- 6 mesi prima lividi frequenti e lenti a scomparire
- Abbassamento dell'emoglobina
- mal di testa e febbre sporadica
- tendance à l'anémie
- pas de symptomes caracterisés
- manque d'appétit, vomissements
- vroeggeboorte 2e kind (onverklaarbaar)
- Kreeg het al op 2-jarige leeftijd
- Hartkloppingen
- Hoge bloeddruk. HELLP

16. Were there any consequences of misdiagnosis; or delay or lack of diagnosis? (Check all apply).

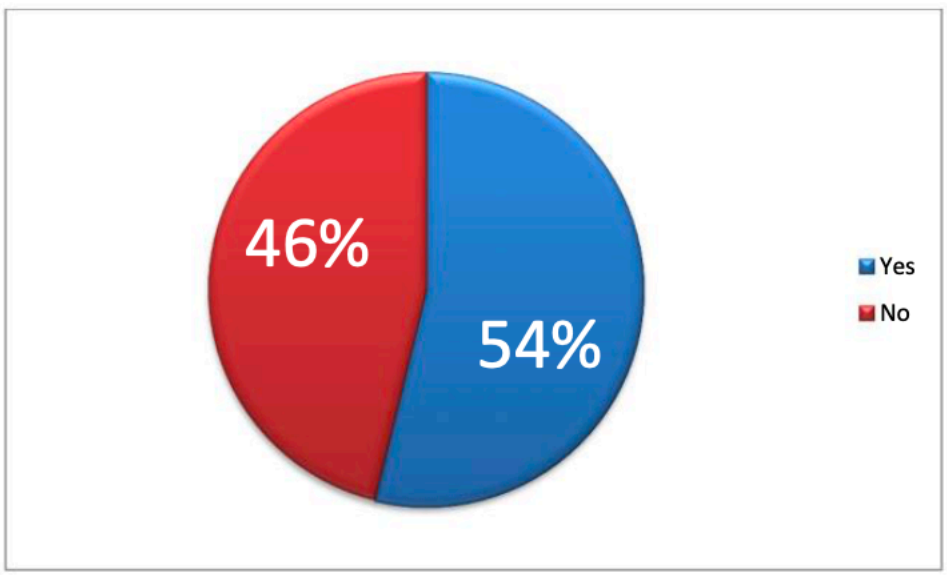


Comments

- Complete kidney failure
- Don't know
- not diagnosed the first time....suffered a relapse exactly 1 year later.
- possible more severe damage generally, but in this case, "assumed" aHUS was treated quickly pending formal genetic testing
- Extreme high blood pressure leading to multiple seizures and 5 days in ICU on ventilator, no diagnosis of aHUS until 8/9 months AFTER this episode, were told this admission to hospital was due - to a 'rare reaction to the dialysis' and 'would never happen again' - it did within a few days of transplant 8/9 months later
- Transplant almost failed 18 months after original kidney failure because of aHUS which was when it - was finally diagnosed. I was also extremely ill at this time. There were also seizures six months after I first became ill which can now be explained by the aHUS diagnosis
- treated even >5y as for TTP, but no aHUS-specific tx at the time
- Too much time spent with plasma pheresis!
- they wanted to remove my kidneys for transplant, they removed 1, then we went to another hospital, - lucky for me ;)
- Dialysis, bleed on occipital lobe seizures
- Lack of confidence in Dr
- Kidney failure
- unnecessary testing of other illness
- Seizures due to high blood pressure
- dialysis, reduced pulmonary function, cardiopathy
- Since our other son had aHUS, this son was diagnosed quickly and was an early adopter of Soliris

- Lesión renal severa
- fracaso renal agudo y entrada en hemodialisis.
- la seu è stata diagnosticata lo stesso giorno in cui il bambino è stato ricoverato. (non si sapeva che era una seu atipica)
- No
- aux urgences pediatriques diagnostic tres rapide
- Kan het me niet herinneren.
- overlijden van ons kind
- Nee, want er werd adequaat direct gestart met plasmaferese en daarna dialyse
- ik ben nierpatiënt geworden (dialyse) maar dat komt niet door de arts die al op de EHBO HUS constateerde maar door mezelf omdat uk veel te laat naar de dokter toe ben gegaan.

17. Do you have access to a national expert /specialist centre for atypical HUS?

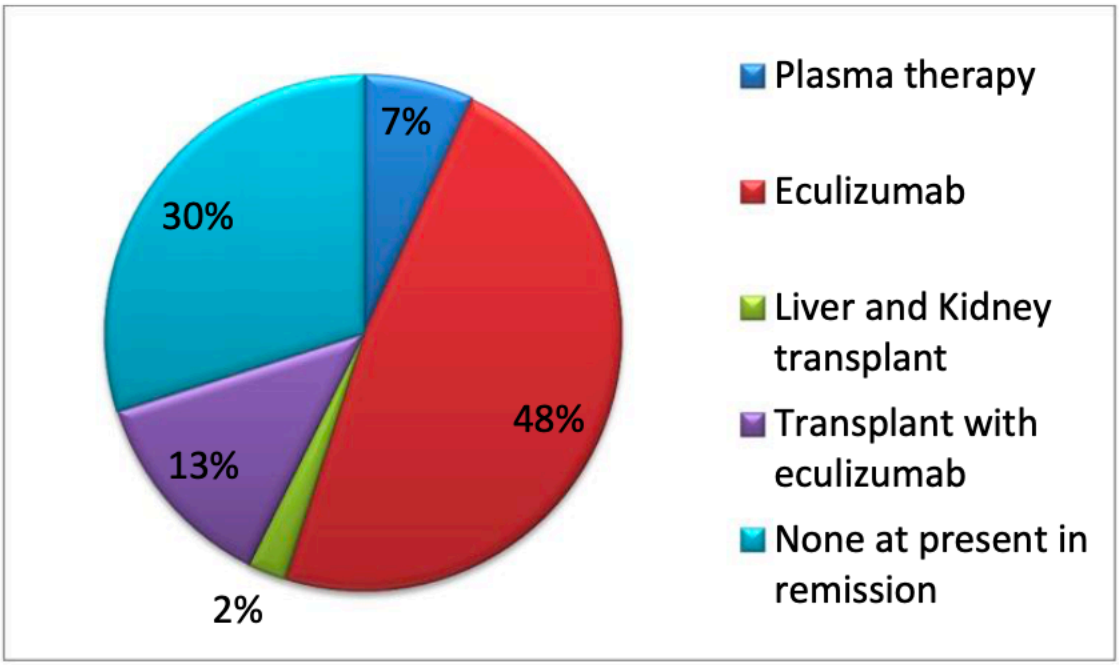


* No response 4

Where? (Duplicate entries eliminated from responses):
- Prof Tim Goodship, Newcastle, Tyneside, Freeman Hospital
- University of Iowa Hospital
- Altnagelvin Hospital Derry N.Ireland Prof Goodship
- Exeter, Devon
- University of Pennsylvania, Philadelphia PA USA
- Alder Hey
- Seattle Children's Hospital, United States
- Birmingham children's hospital
- Lund
- Ohio State Medical Center
- Foundation for Children with aHUS
- Pittsburgh, Pa.
- Children's hospital detroit, mi
- denver, colorado
- Johns Hopkins Hospital
- Children's Hospital Colorado Dr. Gary Lum
- montreal children's hospital
- Cincinnati Children's Hospital Medical Center
- How do we get access to an expert/specialist?
- Doernbecher hospital
- Indiana University Simon Cancer Center Dr. Naveen Manchanda
- Dr. Kaplen

- UT Southwestern, Dallas Texas
- Newcastle
- Rochester General/ Dr. Kouides, and Iowa University
- Toronto
- Bergamo
- Children's Healthcare of Atlanta - Egleston
- MORL research lab
- Royal Childrens Hospital Victoria Australia
- HOSPITAL UNIVERSITARIO LA FE VALENCIA
- Hospital de Sant Pau, Vall d'Hebron
- Texas
- HOSPITAL ITALIANO DE BUENOS AIRES
- Valencia, España
- Hospital Clínic, Barcelona
- la fe (valencia)
- Milano clinica de marchi
- Mario Negri di Ranica (Bergamo)
- Clinica Pediatrica De Marchi
- BERGAMO – FIRENZE
- arnaud de Villeneuve Montpellier
- hopital nord d'amiens
- Timone marseille
- Hôpital tenon Paris
- CHU Nantes
- Chu de Rouen
- hopital arnaud de villeneuve
- bruxelles
- Alger
- hopital
- poitiers chu
- Lausanne
- Belgique, Bruxelles, Cliniques St Luc
- Marseille
- CHRU de lille
- UZ Antwerpen
- Nijmegen (RadboudUMC)
- uni klinik köln

18. Current treatment for atypical HUS



* No response 10

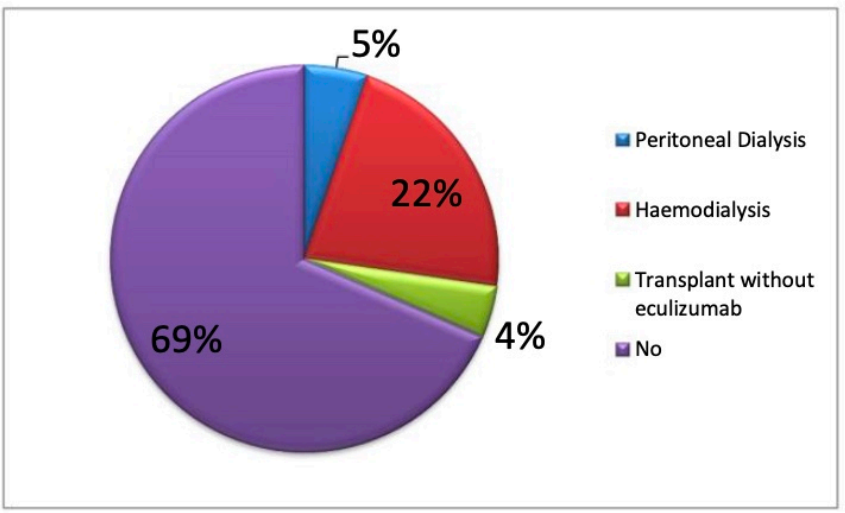
Comments

- failed kidney transplant & awaiting transplant with eculizumab
- dialysis
- Eculizumab (Soliris) infusion every other week and 5 mL penicillin twice daily
- Dialysis
- Waiting for a second kidney transplant with eculizumab
- kidney transplant, with eculuzimab
- On dialysis
- Kidney transplant / HUS in remission
- On daily Hemodialysis
- only medication for bloodpressure and every months kidneytests
- Steroids and anti rejection meds
- Soliris infusions
- Trying to locate live donor for transplantation
- on dialysis waiting for transplant
- on dialysis
- Hemodialysis, Blood Pressure meds, EPO, Vitamin D, Iron
- following kidney transplant - prior to transplant treated wtih IVIG, plasma exchange, pla infusions and then peritoneal dialysis once he reached ESRD
- Steroids, Ferrosig. Mircera, BP medication
- steroids, high blood pressure tablets, Azathioprine
- tratamiento con cellcep, trasplante con 9 dosis de inmunoglobuluna y belataced.
- in fase di rivalutazione terapeutica
- DIALISI
- aucun actuellement car plus de poussées vu que je n'ai pas de reins
- Arrêt de eculizumab depuis septembre 2013 après 50 cures
- au début endoxan, puis corticoïdes, puis hydrocortizone
- greffes renale sans eculizumab
- sous dialyse
- Maandelijkse bloedcontrole na stop Eculizumab.
- Ben 2.5 jaar geleden getransplanteerd, HUS is rustig.
- getransplanteerd
- niks
- Wacht op transplantatie met Eculizumab
- in 2000 getransplanteerd, daarvoor peritoneale dialyse

Living with Atypi
Syndrome Poll R

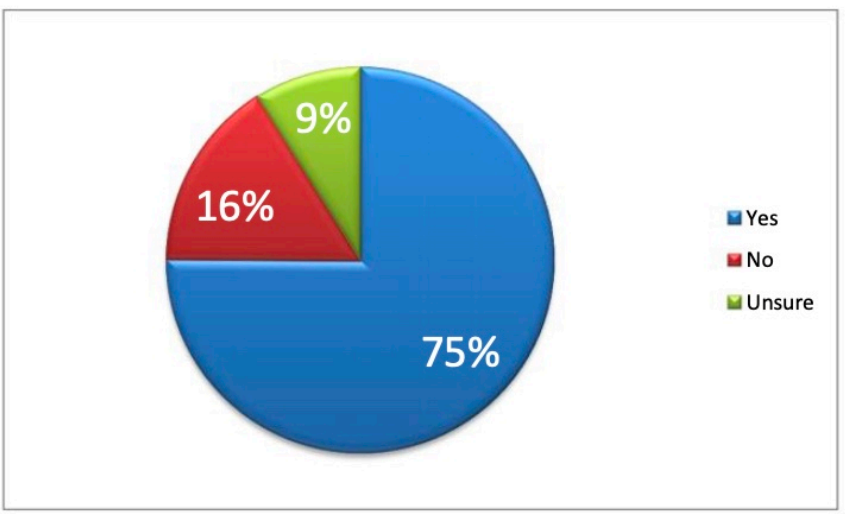
- Wel gebruik ik een groot aantal bloeddrukverlagende medicijnen, EPO en een bloedverdunner
- dit moet nog gaan gebeuren
- in remissie dmv medicijnen

19. Are you receiving renal replacement therapy?



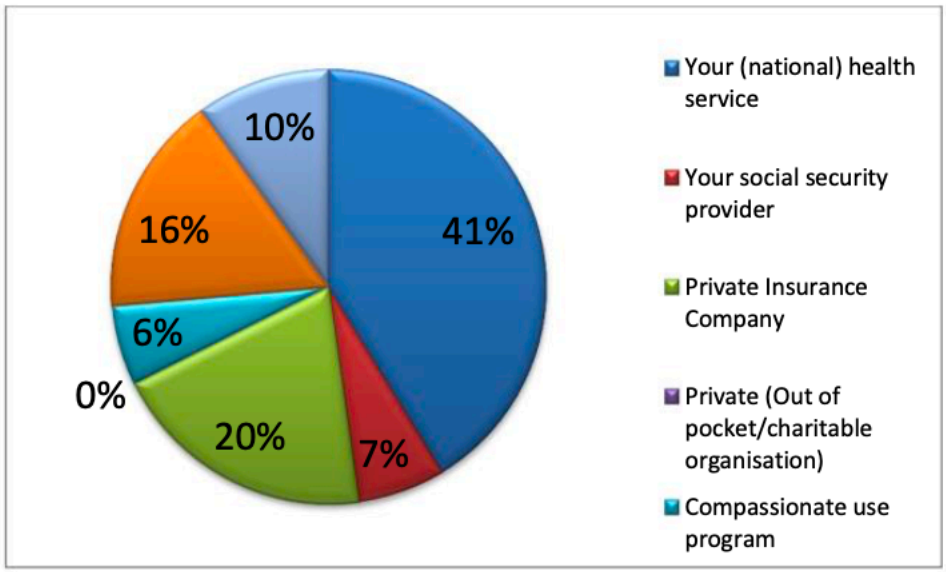
*No response 5

20. Is your current treatment your treatment of choice?



*No response 1

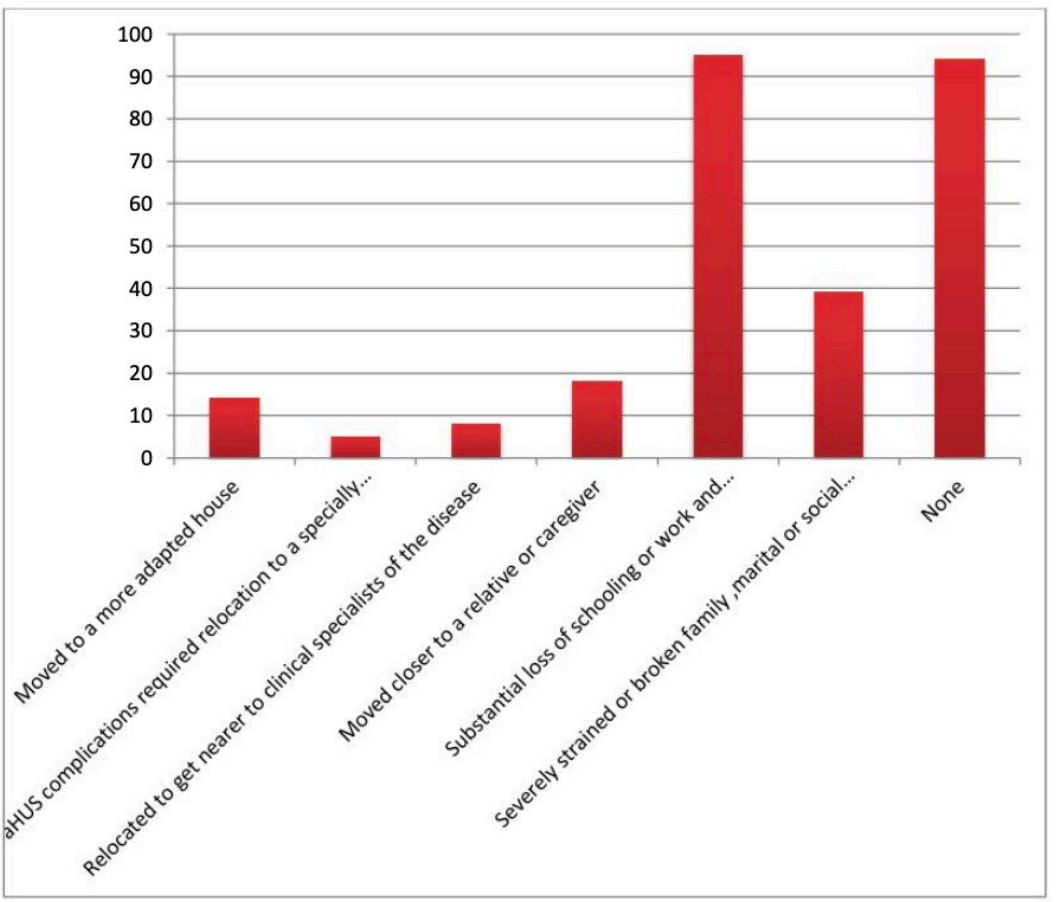
21. **If you have access to eculizumab treatment, who will pay for it?**



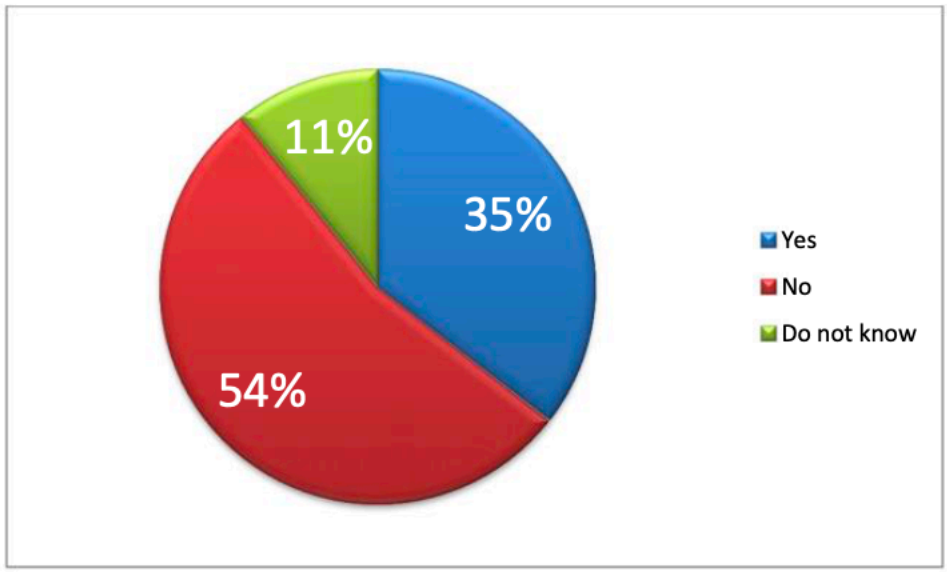
*No response 13

Other (Please specify)
- Not sure if nhs in uk supports this drug treatment.
- unsure - local PCT on "one off" "temporary" basis?
- Alexion at the moment
- A combination of Private Insurance, our own income, and patient assistance program from - Alexion (drug company)
- Consultant would appeal if needed
- not sure. not interested at time it was offered
- Hopefully hospital
- Currently SS with private insurance taking over after 36 months following transplant
- I don't know if she would qualify for coverage with Medicare
- NYSCHP
- según el médico especialista no lo requiero.
- En este momento no se me esta administrando el tratamiento que necesito para mi enfermedad y así poder ser trasplantada
- OBRA SOCIAL
- NO LO SÉ
- je ne suis pas encore sur la liste d'attente de greffe
- Durant la cure protocole alexion
- pas de traitement pour l'instant
- Weet niet of ik toegang heb
- heb geen idee wat dat is
- nooit van gehoord

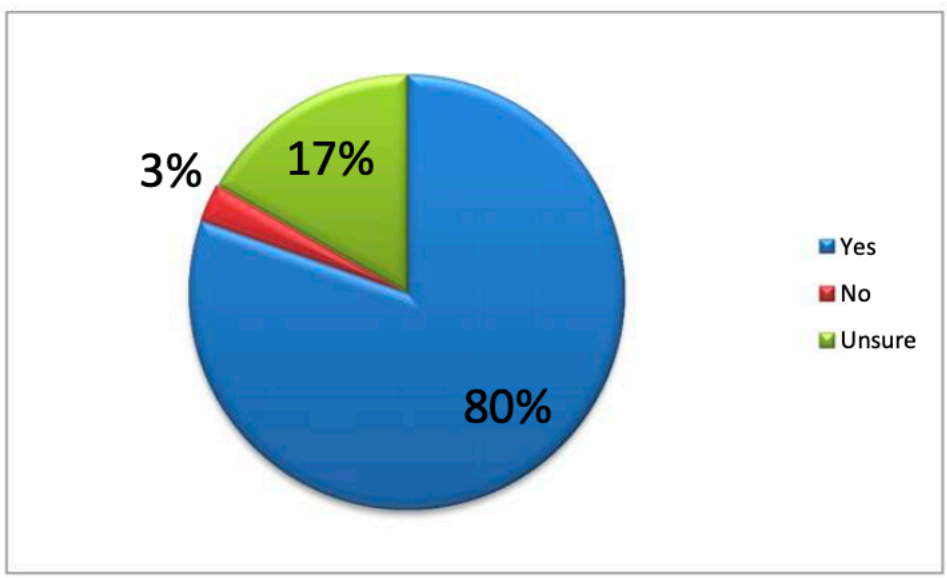
22. **Did the disease cause changes in housing or lifestyle? (please check all that apply)**



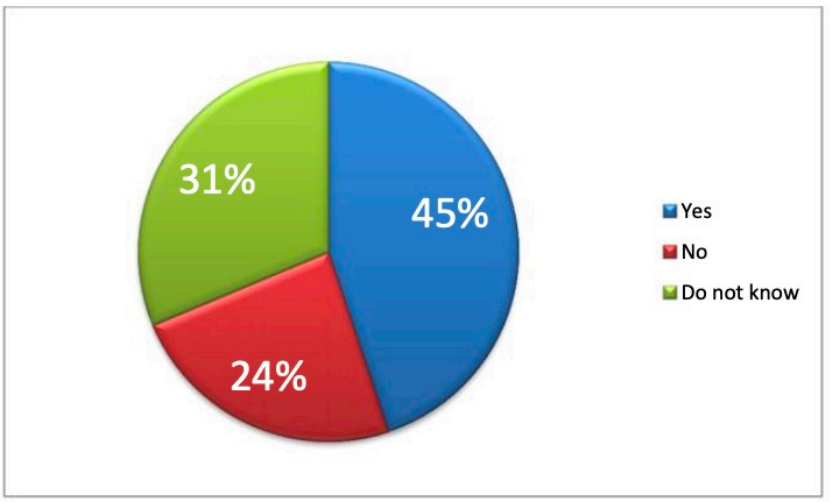
23. **Have you participated in research into aHUS in the past?**



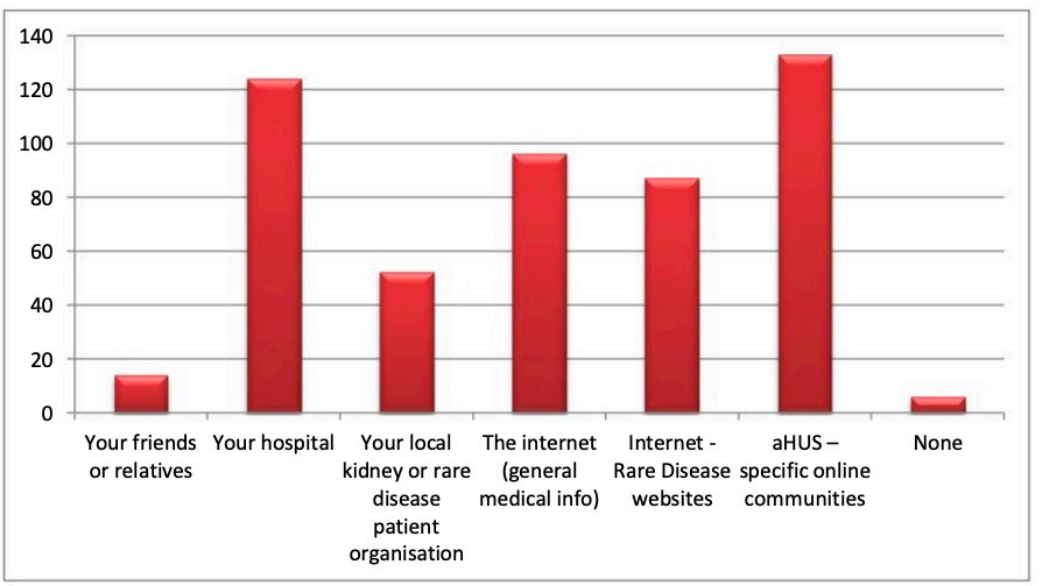
24. **If you had the opportunity would you be prepared to participate in research about atypical HUS in the future including full genome (DNA) sequencing?**



25. **Are you enrolled in an atypical HUS clinical registry for research?**

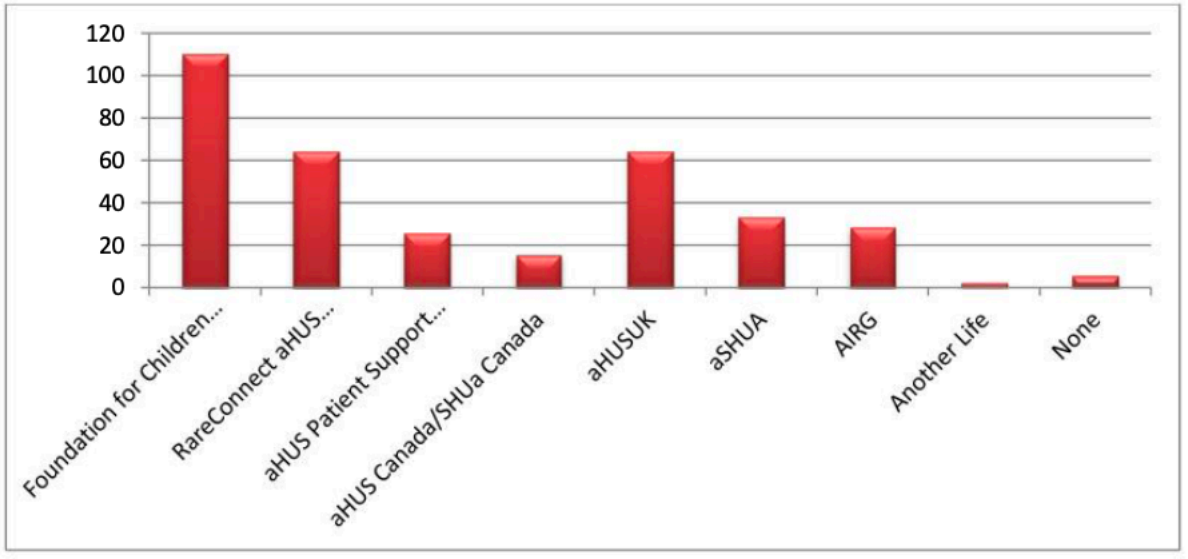


26. **From what sources do you receive information about atypical HUS? (check all that apply)**



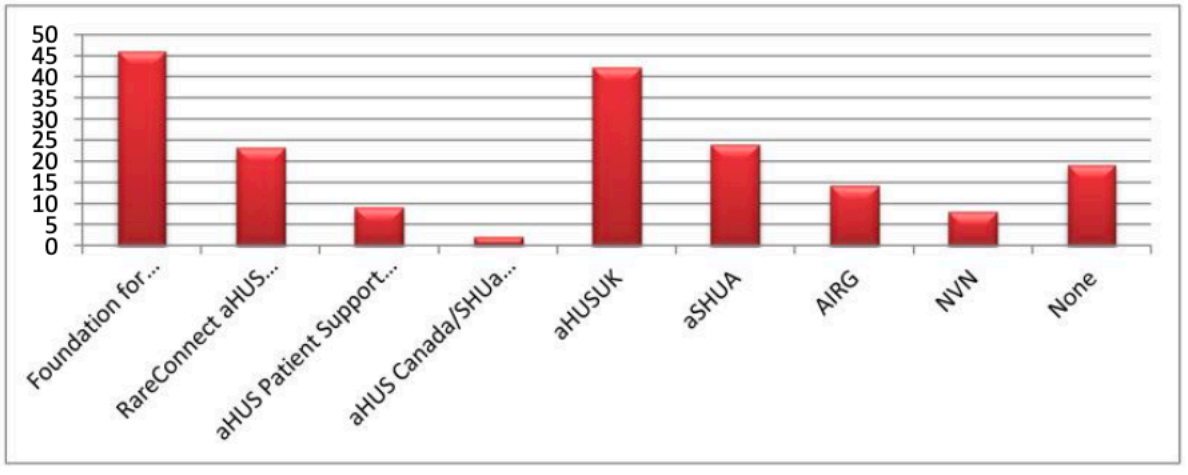
*No response 3

27. **Prior to taking this survey which of the following patient organisations were you aware of? (check all that apply)**



*No response 5

28. Are you a member of any of the following atypical HUS patient groups?´



*No response 17

Other

- I don't know if I'm a member.
- apec (argentina)