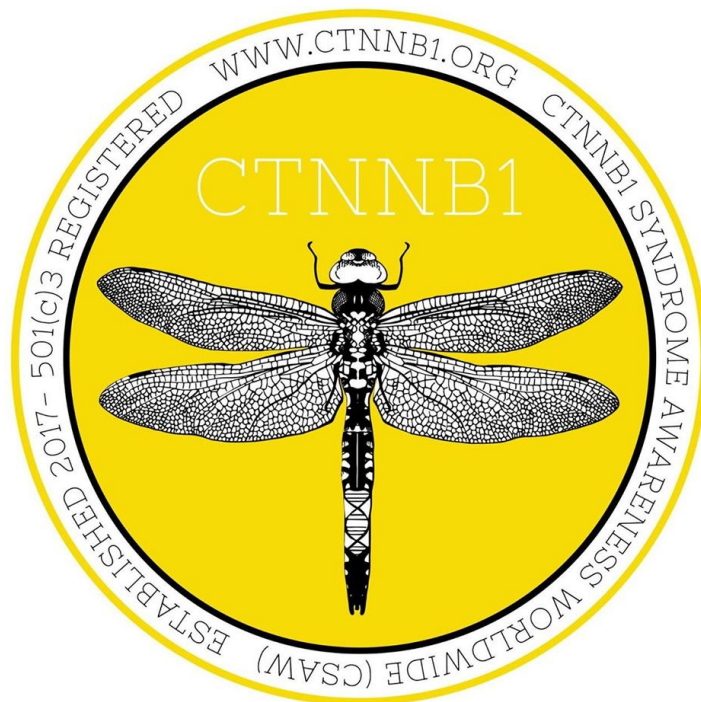
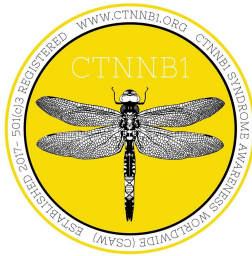


CTNNB1 SYNDROME

Survey Annual Report



A RESOURCE FOR FAMILIES AND PHYSICIANS
SEPTEMBER 2020



CTNNB1.org sponsors a comprehensive survey of those with CTNNB1 Syndrome to aggregate information on the disorder. This report compiles the data of those who have completed the survey. It is offered as a resource for parents, doctors, therapists, and researchers to know what to expect upon diagnosis of a CTNNB1 syndrome diagnosis, and what are some common impacts, symptoms, behaviors, and therapies that have proved effective for some, and more.

If you have a child with CTNNB1 Syndrome, please give strong consideration to completing this survey and signing up for Simons Searchlight to help other parents and the medical community learn about the experience of CTNNB1 children and families.

PARTICIPATE HERE:

CTNNB1 SURVEY:

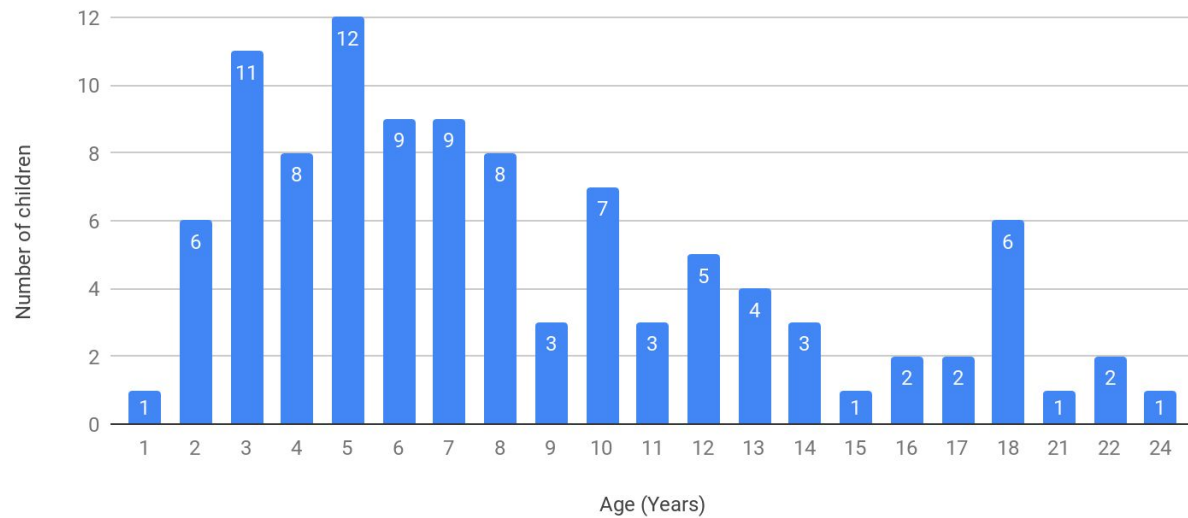
<https://www.ctnnb1.org/registry>

SIMON'S SEARCHLIGHT:

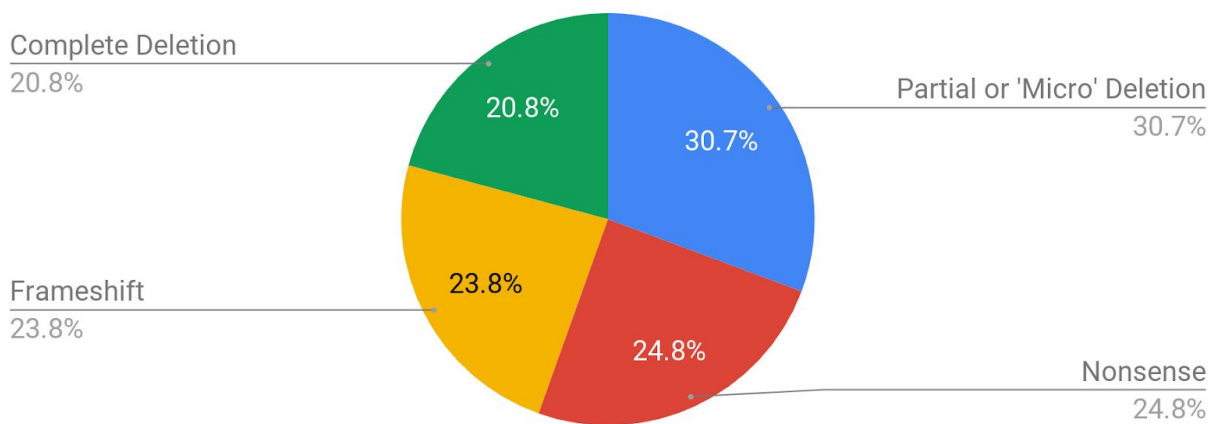
<https://www.curectnnb1.org/natural-history-study>

Thanks to CTNNB1 volunteer Bruno Ramalho for his outstanding work on creating this valuable and user-friendly resource for our families.

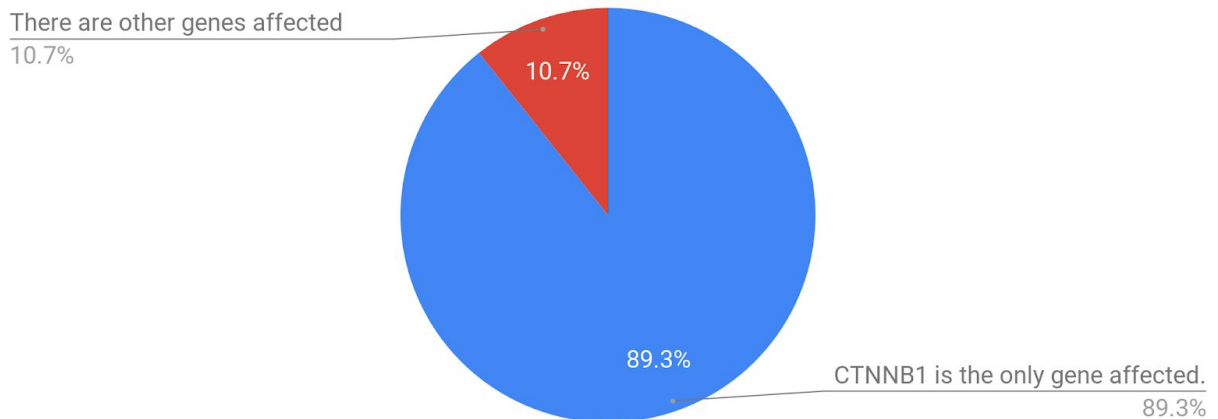
Age Distribution as of July 2020 (104 children)



Please indicate the extent of the deletion or mutation of the CTNNB1 gene.



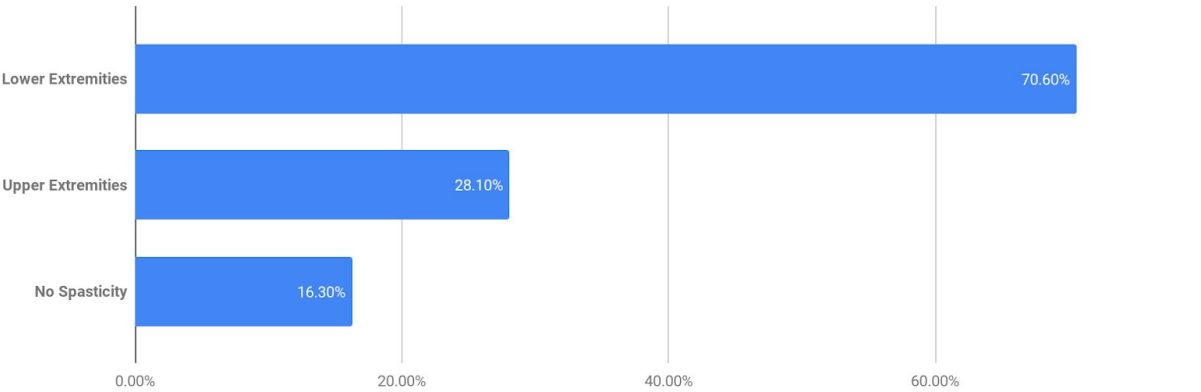
Is the CTNNB1 the ONLY gene that is affected in the subject?



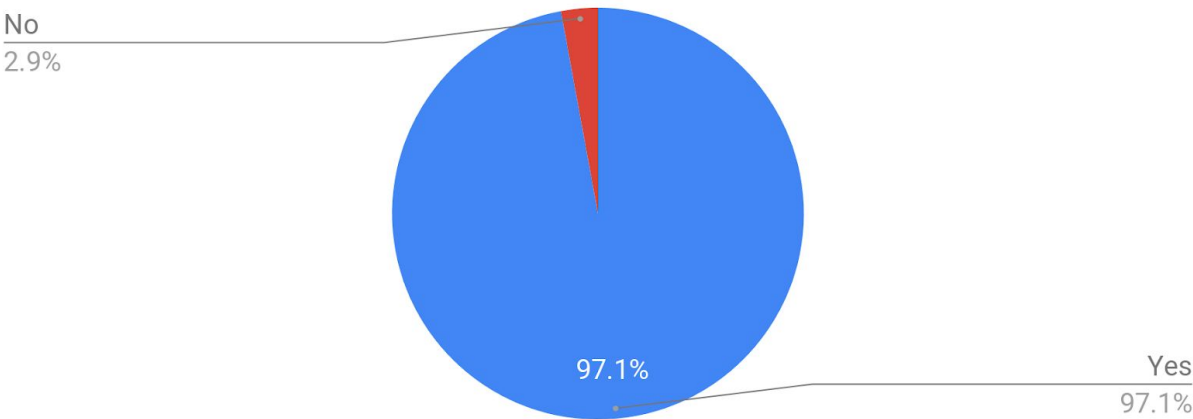
Other genes that are affected

GLA GENE INHERITED FROM HER FATHER aka FABRY DISEASE	1
22q11.2 and unbalanced Translokation 16/19	1
Men2a	1
HUWB1	1
EP300	1
Ulk4	1
AFF4	1
ULK4, TRAK1, CCK, LYZL4, VIPR1, SEC22C,SS18L2, NKTR, KLHL40, HHATL,, CCDC13, HIGD1A,, ACKR2, KRBOX1, CYP8B1,, ZNF662, FAM18A, POMGNT2, SNRK, ANO10, ABHD5, ZBTB47.	1
NM_001904.4:c.735-1G>C p ? (MIM 615075)	1
Brca1	1
kansl1, mbd5, klf1a, ndufa6, dnmt3b, sobp	1

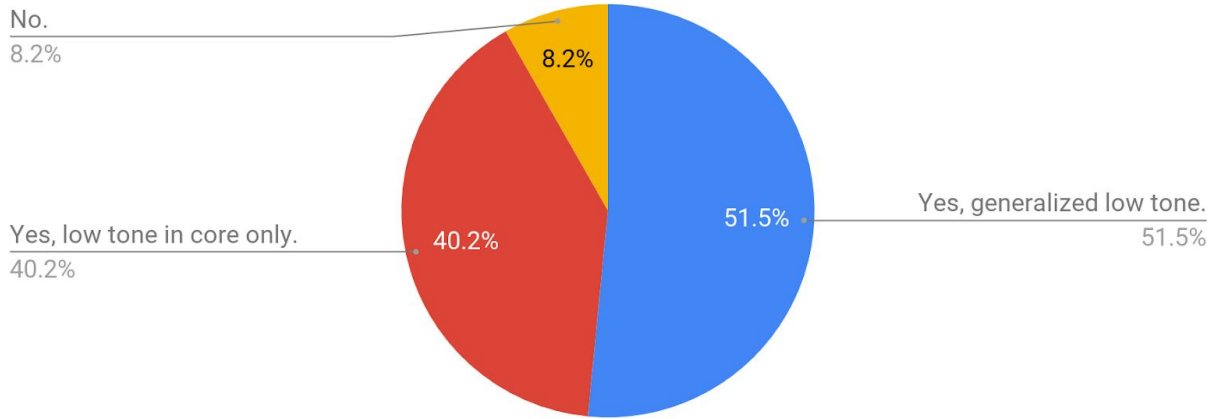
Does the subject have spasticity? If yes, where?



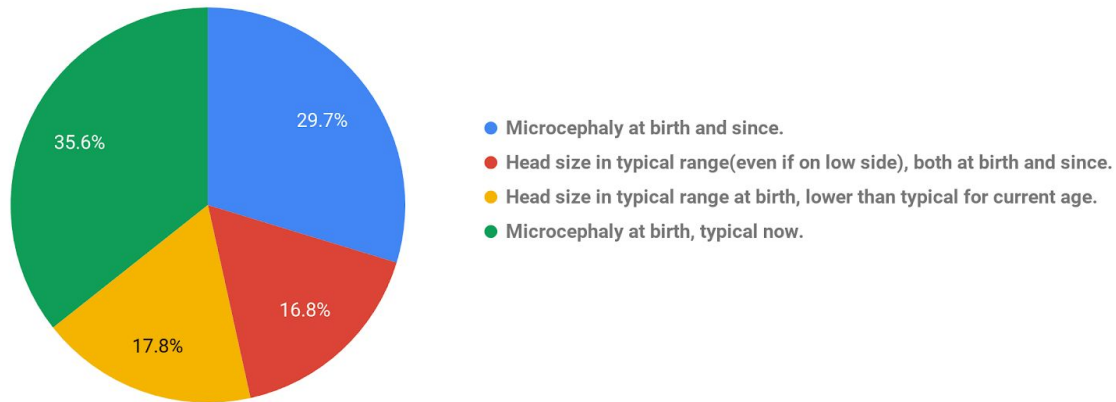
Did the subject present with global developmental delay?



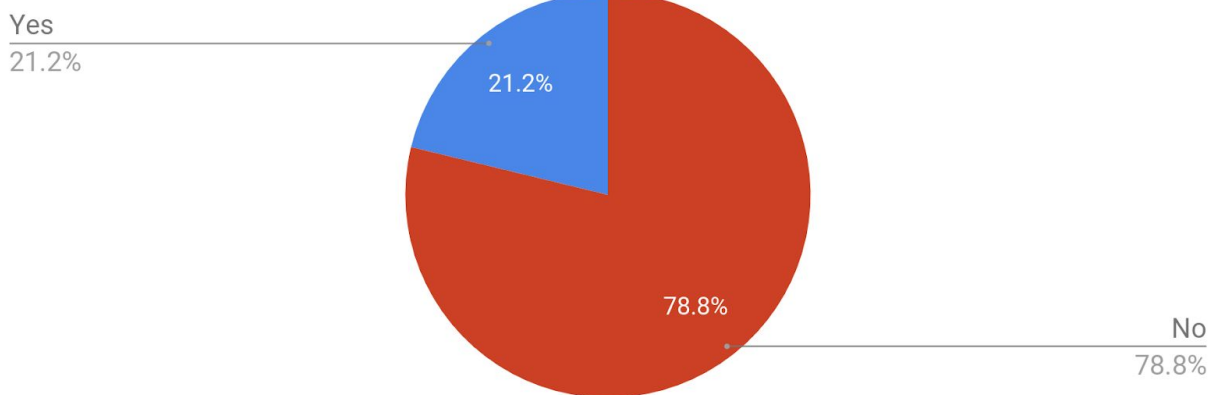
Did the subject present with hypotonia (low muscle tone)?



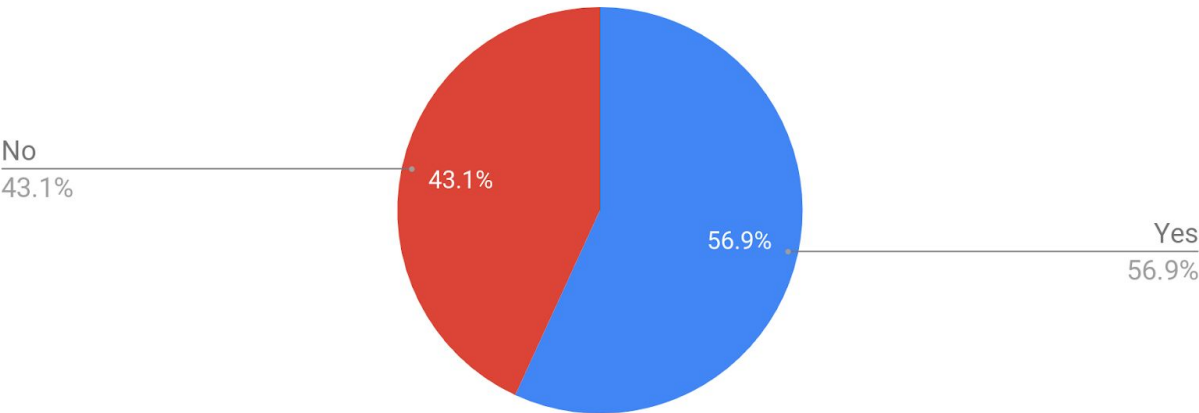
Did the subject present with small diameter head or microcephaly at birth?



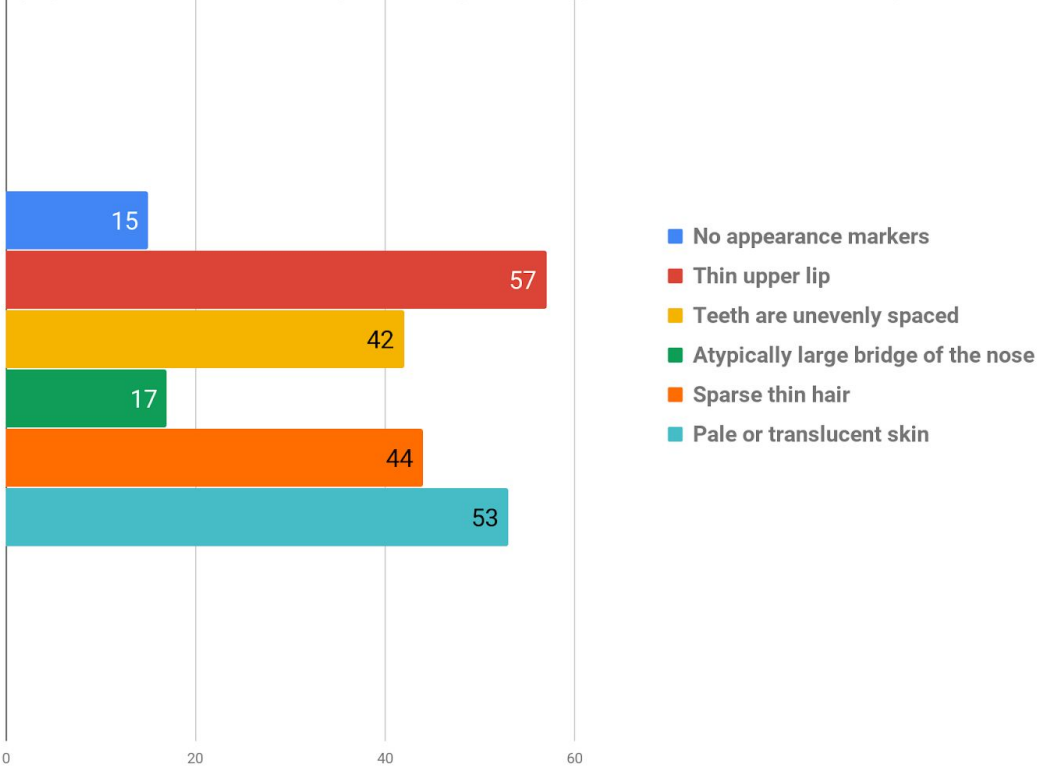
Has the subject suffered a bone fracture?



Has the subject had issues with congestion, swallowing or choking?



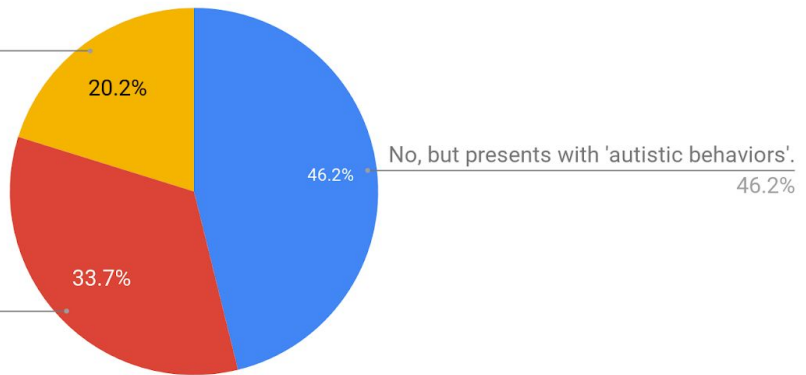
Symptoms: Which of the following CTNNB1 Syndrome 'appearance markers' does the subject have?



Has the subject been diagnosed with Autism Spectrum Disorder, or present with 'autistic behaviors'?

Yes, diagnosed.
20.2%

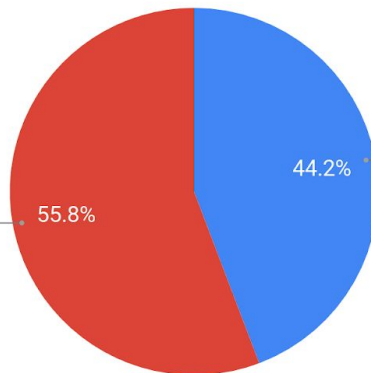
No
33.7%



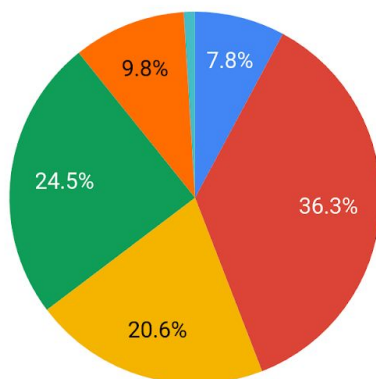
Has the subject been diagnosed with sensory processing issues?

No
55.8%

Yes
44.2%

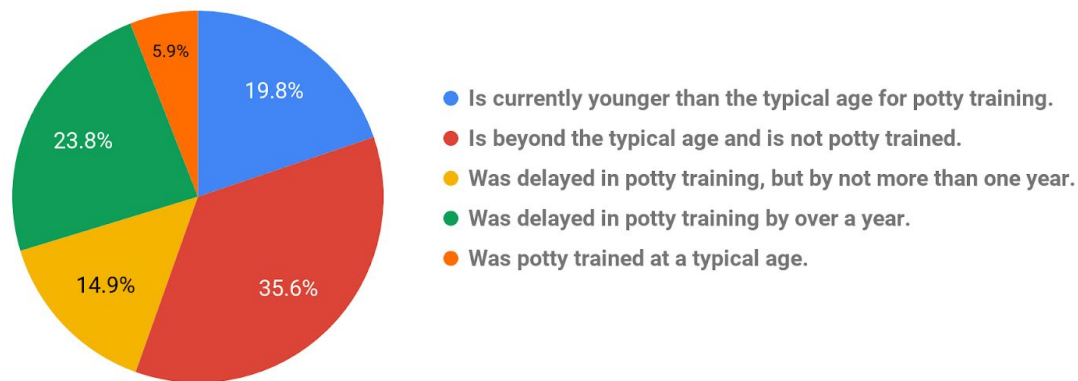


If the subject is at or beyond the 'typical' age for walking unassisted, select the most relevant statement.

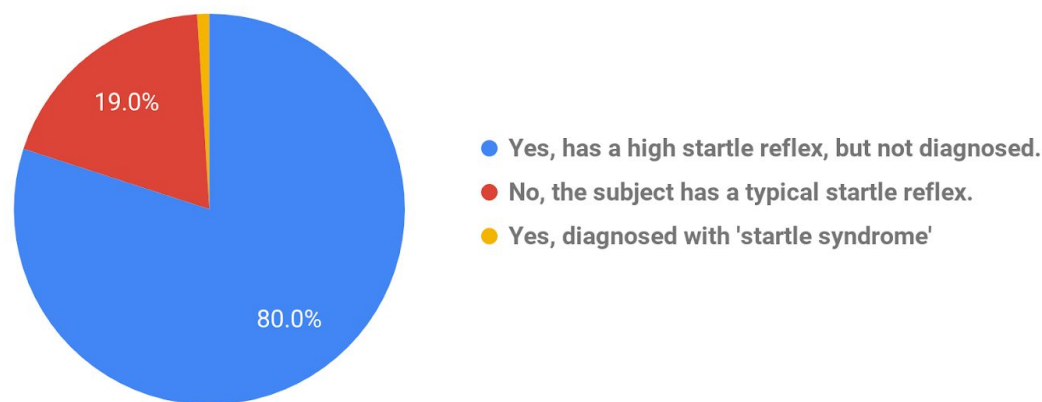


- Is currently younger than the typical age for walking.
- Was delayed in walking by over a year.
- Walks with the assistance of a walker (but was delayed).
- Is beyond the typical age and is not walking.
- Was delayed in walking, but by not more than one year.
- Walked at a typical age.

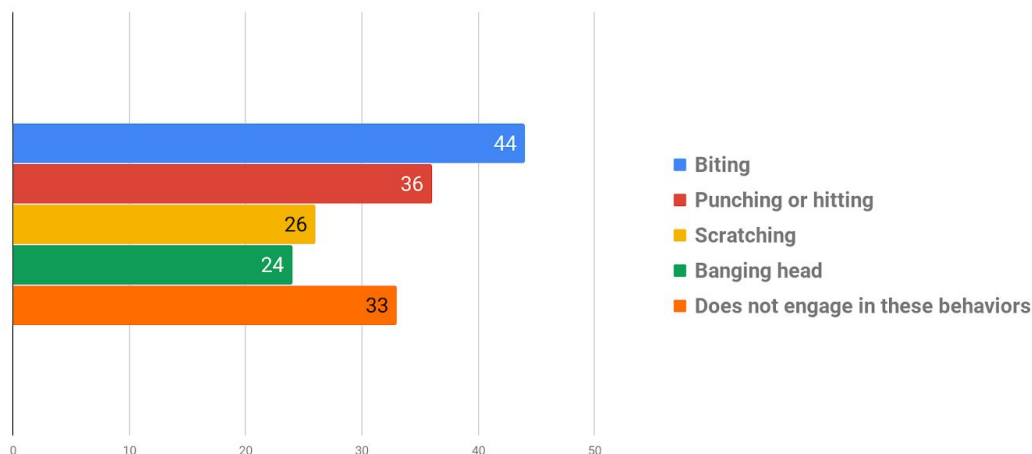
If the subject is at or beyond the 'typical' age for potty training, select the most relevant statement.



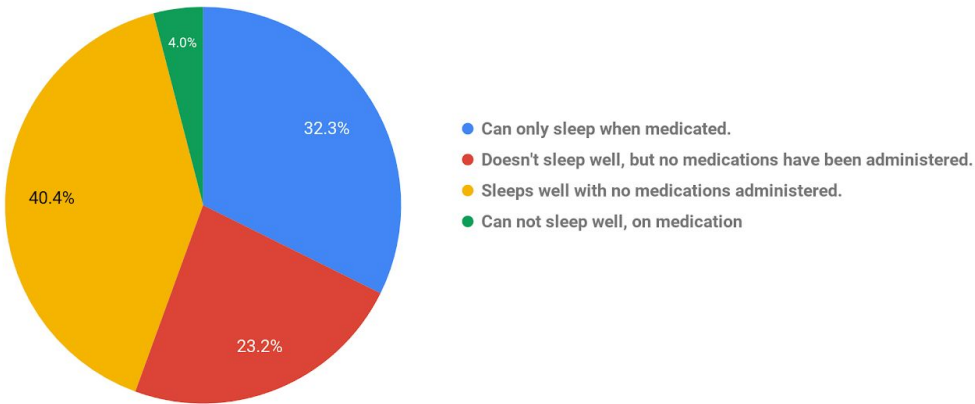
Has the subject been diagnosed with 'startle syndrome' or does the subject have a high startle reflex?



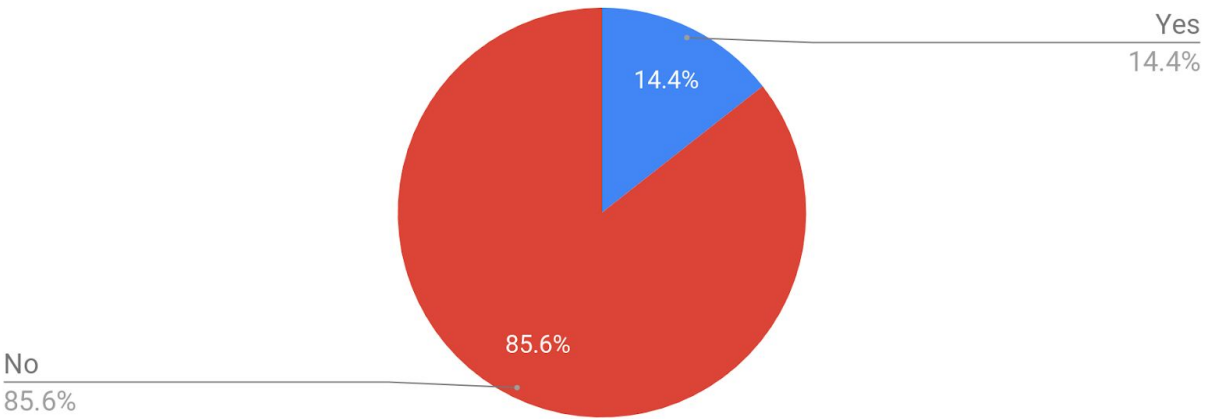
Does the subject engage in self-harm or injurious behavior to others?



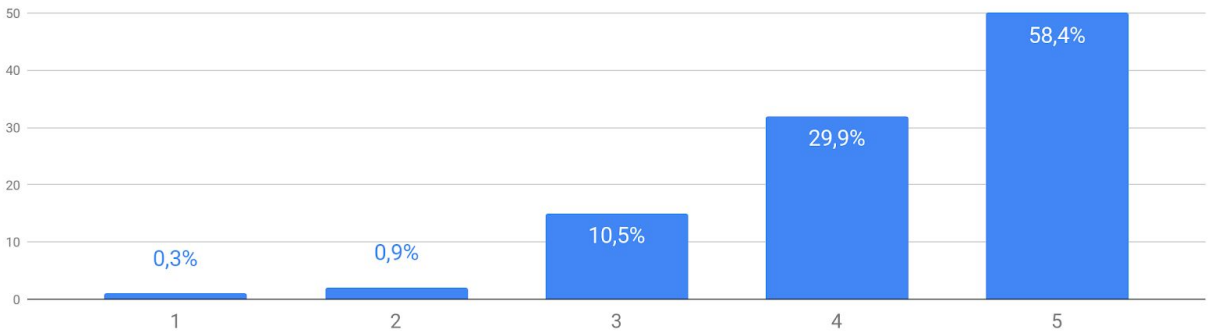
Please indicate the subject's quality of sleep:



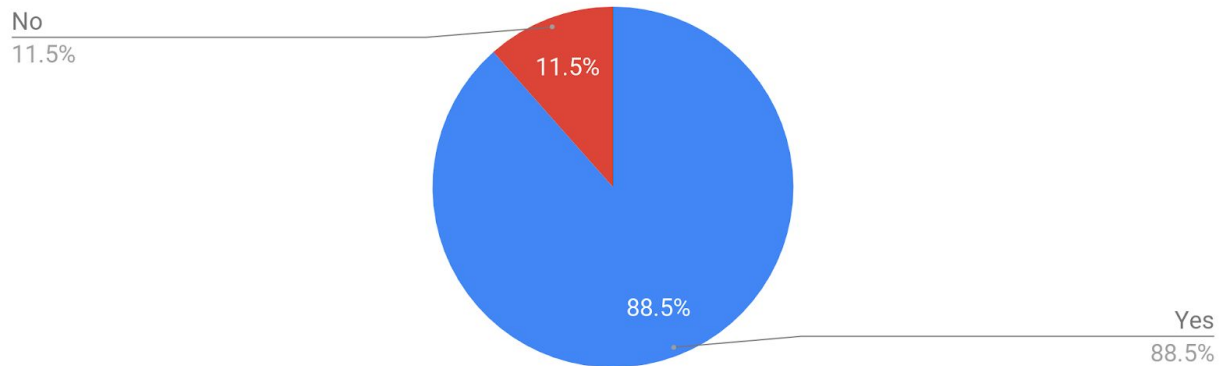
Has the subject been diagnosed with any heart issues?



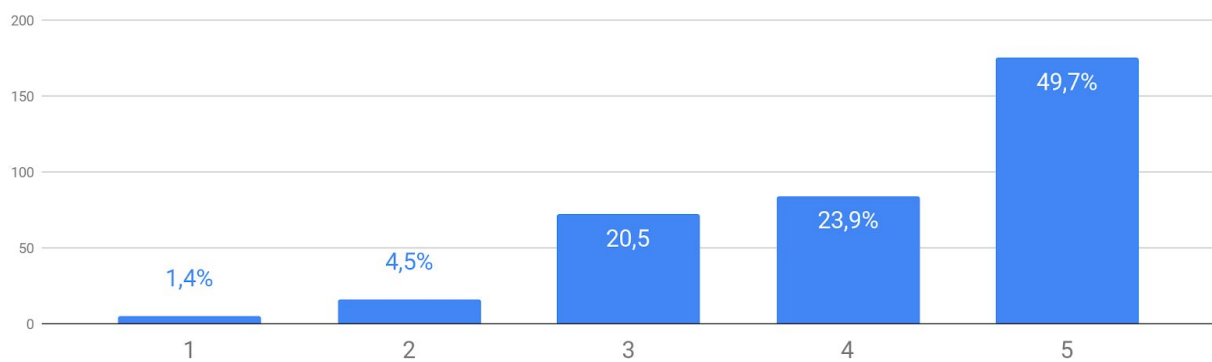
If the subject has participated in physiotherapy/physical therapy, how would you rate the effectiveness of this therapy?



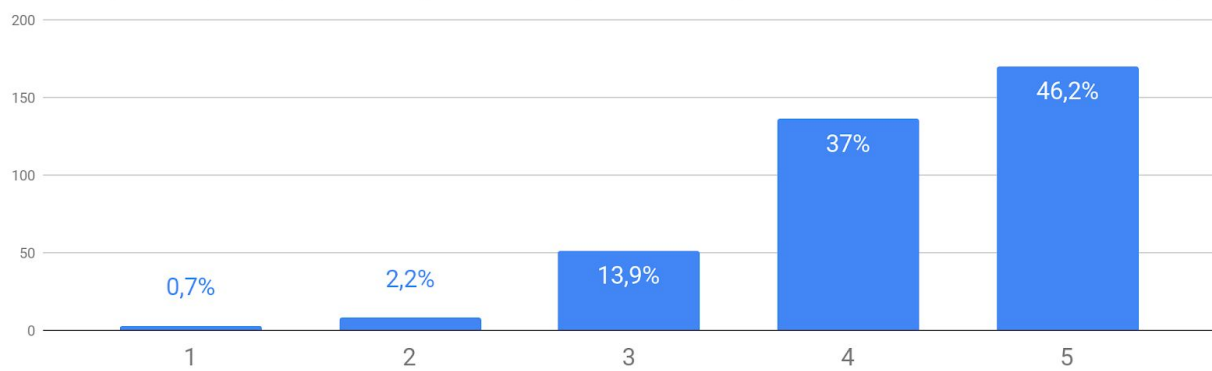
Has the subject participated in speech/language therapy?



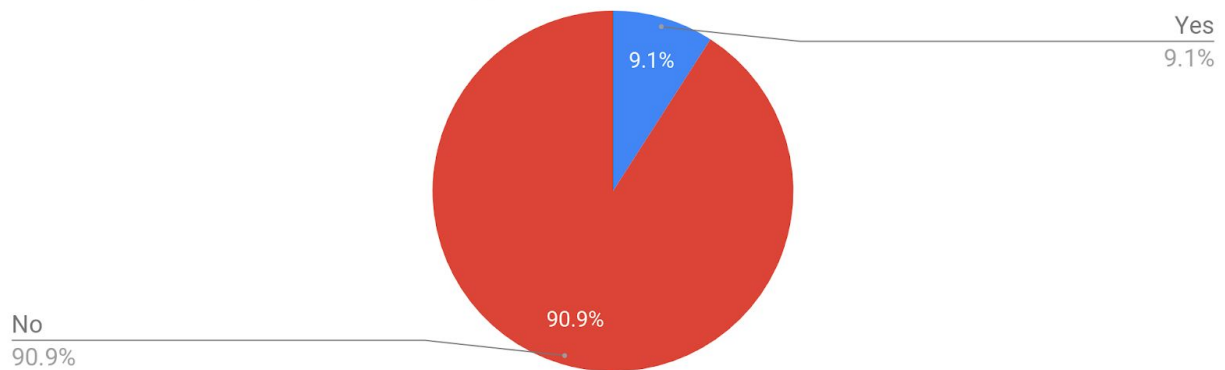
If the subject has participated in speech/language therapy, how would you rate the effectiveness of this therapy.



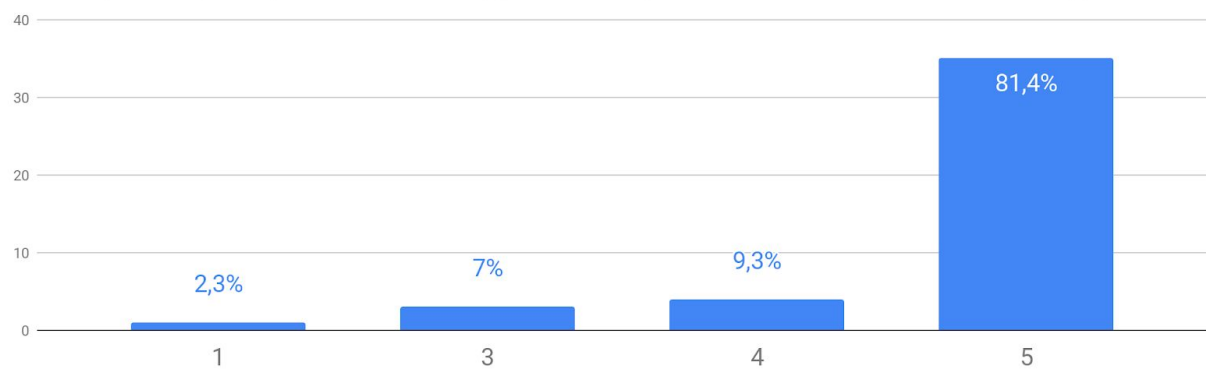
If the subject has participated in occupational therapy, how would you rate the effectiveness of this therapy?



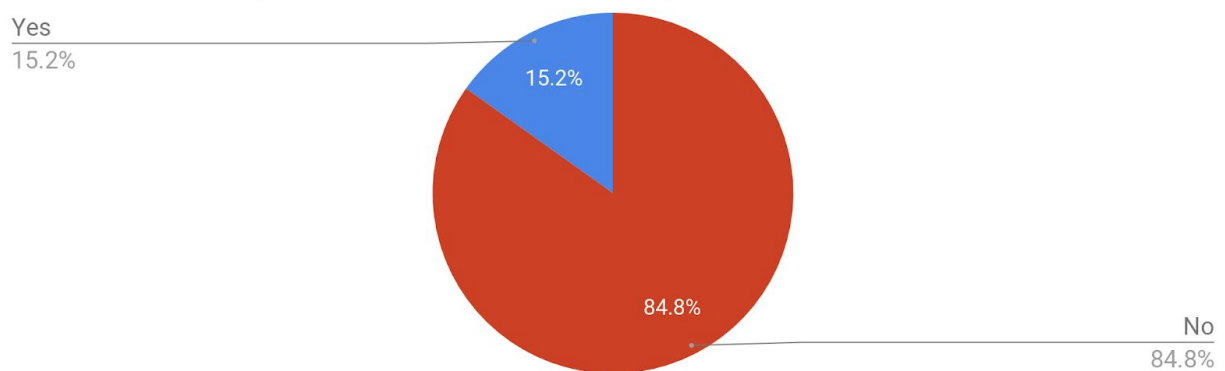
Has the subject participated in ABA Therapy?



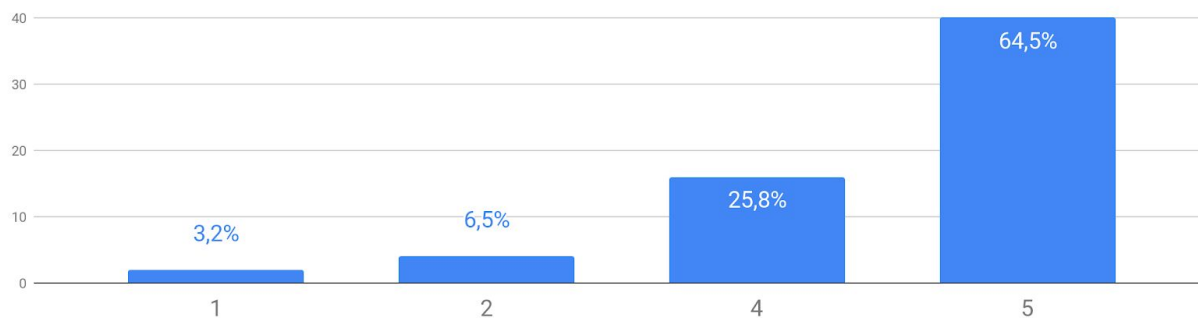
If the subject has participated in ABA Therapy, how would you rate the effectiveness of this therapy?



Has the subject participated in Conductive Education Therapy?



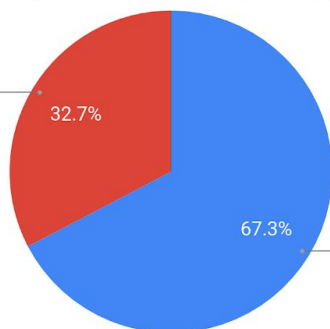
If the subject has participated in Conductive Education Therapy, how would you rate the effectiveness of this therapy?



Has the subject participated in hippotherapy (equine/horseback riding therapy)?

Yes

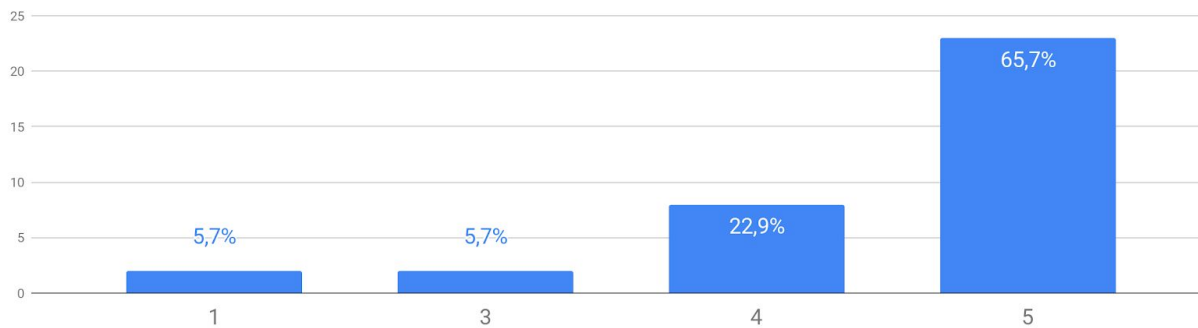
32.7%



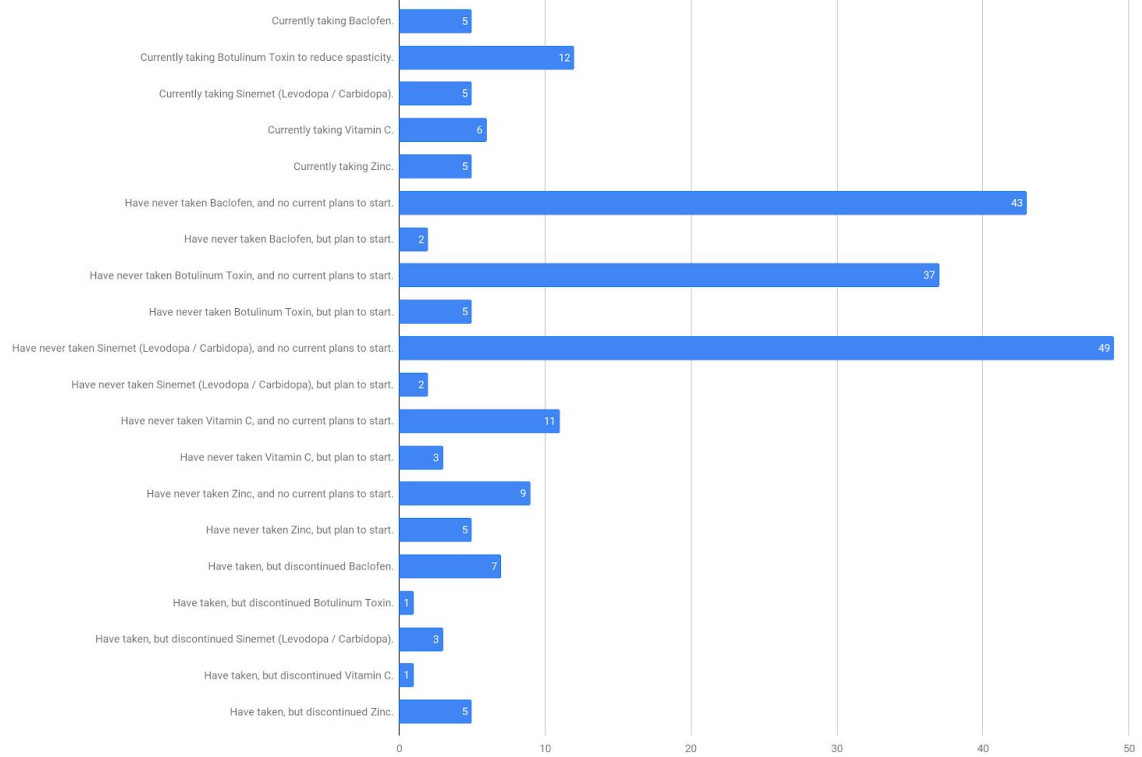
No

67.3%

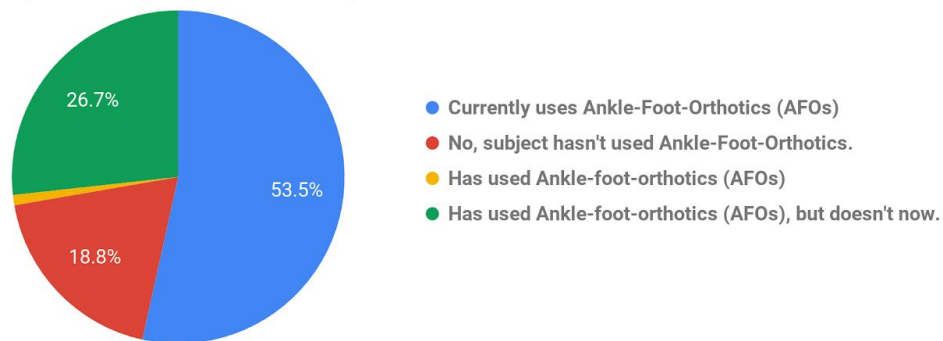
If the subject has participated in hippiotherapy, how would you rate the effectiveness of this therapy?



Which of the following best describes the drug therapies that the subject has undergone in relation to CTNNB1 Syndrome:



Does the subject currently use, or has the subject used Ankle-Foot-Orthotics?



Does the subject currently use, or has the subject used TheraTogs(c) or Benix(c) shorts for hypotonia?

