

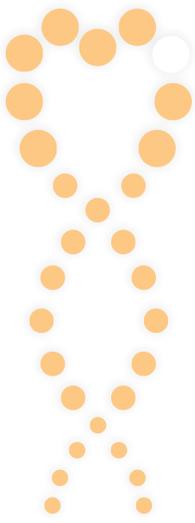


WSS Foundation

Wiedemann-Steiner Syndrome Foundation

CoRDS Registry Data

April 2018



What is CoRDS?

Do you remember the feeling you had when first receiving the diagnosis of Wiedemann-Steiner Syndrome (WSS). After months and years of doctor visits, poking, prodding, scanning, time off from work, time away from family, along with all those parking fees and gas money; I am sure there was initially a bit of elation if you were like me. We finally have an answer! This immediately followed by the fear of the unknown. When Maria and I received Dylan's diagnosis five years ago, the fear of the unknown was overwhelming, and the medical professionals were no help. By pure luck we were one of two families diagnosed by the genetics team at Minneapolis Children's

Hospital. They made us aware of a very small Facebook group consisting of about 15 families at that time. While it was great to be able to connect initially with others that were in the same boat as us, there was no medical data, direction, insight, what-so-ever, available for anyone. The fear of the unknown continued. What was in store for Dylan, what can we expect, WHAT DO WE DO?

Fast-forward to today. There are now hundreds of diagnosed cases around the world meaning we have hundreds of data points to tap for experience and insight. While we, as family members, have the support from the WSS Foundation and separate branches in Facebook groups, local gatherings and the semi-annual WSS Foundation meeting. Now, in a similar way that we have been providing support to each other, we are able to pay it forward in the form of data shared with (to the health care and educational professionals) that give so much back to our children.

Enter CoRDS. CoRDS is a centralized international registry setup by Sanford Health. Think of it as a database for all the details that make our kids special. Everything from GI to glasses, muscle tone to behavioral challenges; all these details are entered in to this completely confidential registry. This provides all your healthcare providers, educational professionals and researchers access to data from the potential totality of the known WSS population.

Imagine if you were able to have all the questions you had flood into your head in the moments, days, and months after receiving the WSS diagnosis answered. With your help of entering your data into the registry, this can be a reality for future diagnosed families. Every data point entered into CoRDS provides an additional breadcrumb for the professionals to learn from. The more data available to those providing care, the better care and direction they are able to provide us.

Please, think back again to when you received your initial diagnosis and ask yourself if a few minutes of your time is worth paying it forward to the next diagnosed and the rest of the WSS community. We ask that you take the time (about 45 minutes, beginning to end) to register [HERE](#) and complete as much info as you can.

While our goal is 100% participation, we have set a goal of 100 registrations before the end of 2018. Keep checking back on the WSS Foundation CoRDS webpage for updates on our achievements and graphs showing some of the unique yet overwhelmingly similar attributes of our kids. We will be updating this data on a semi-annual basis. Additionally, as more WSS specific questions are added to the questionnaire, we will push that info out to you asking that you add to this knowledge base; ultimately making a better world for everyone involved in the WSS community.

Jacob Cummings, *Dylan's Father*
WSS Foundation Board Member

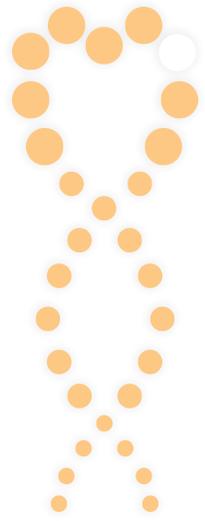
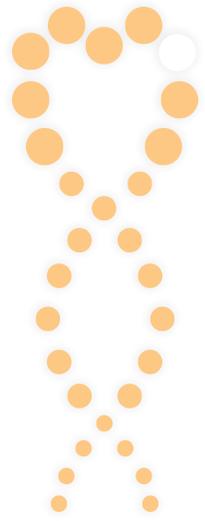
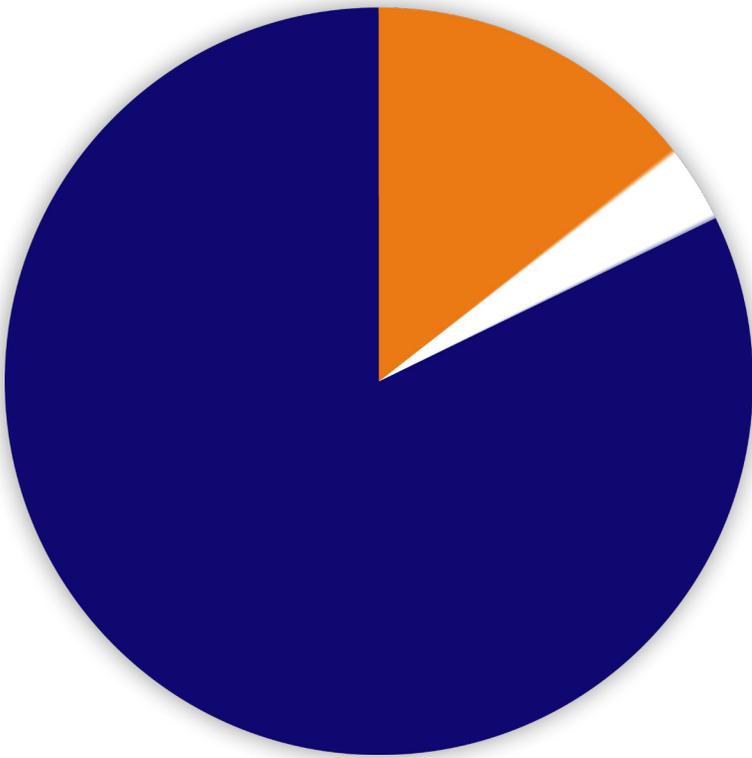


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Growth Hormone Therapy



Key

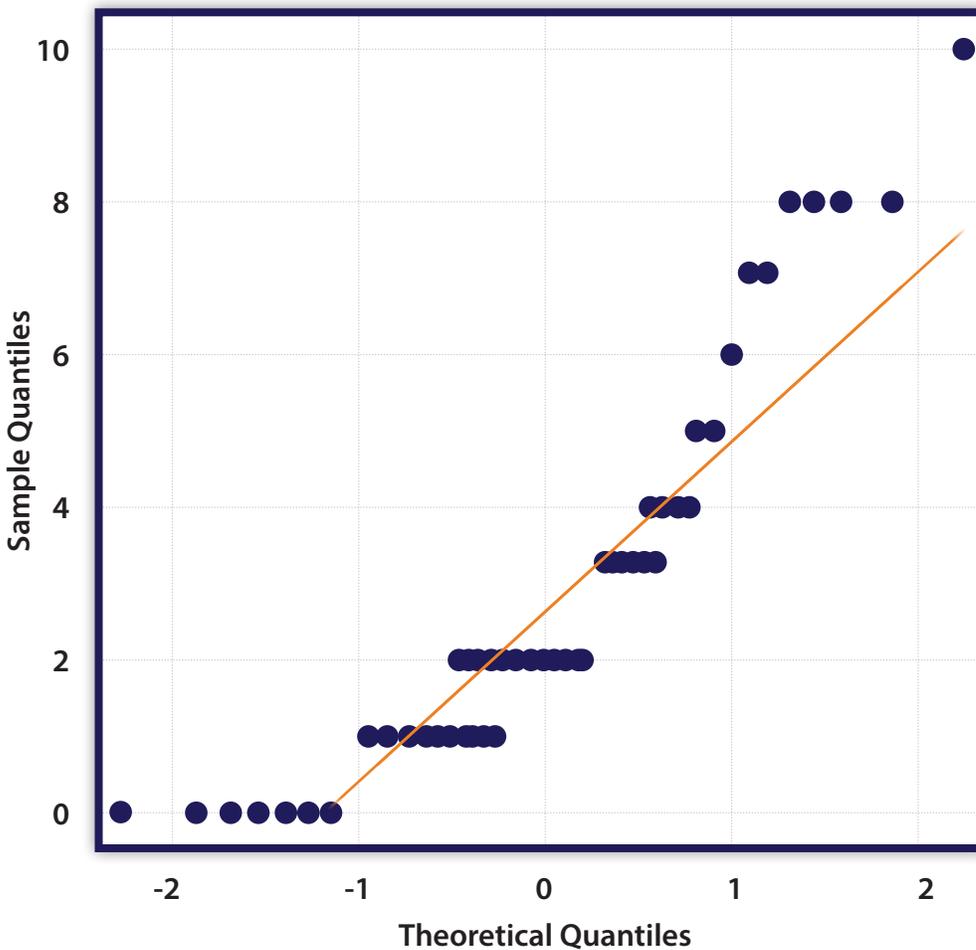
- No
- Yes
- No Information

Notes

Of the respondents, 11% indicated use of human growth hormones.

Short stature is a diagnostic feature of WSS.

Normal Quantile Plot | Number of Surgeries

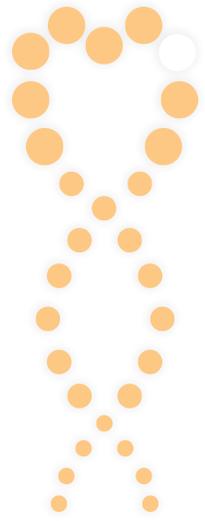


Notes

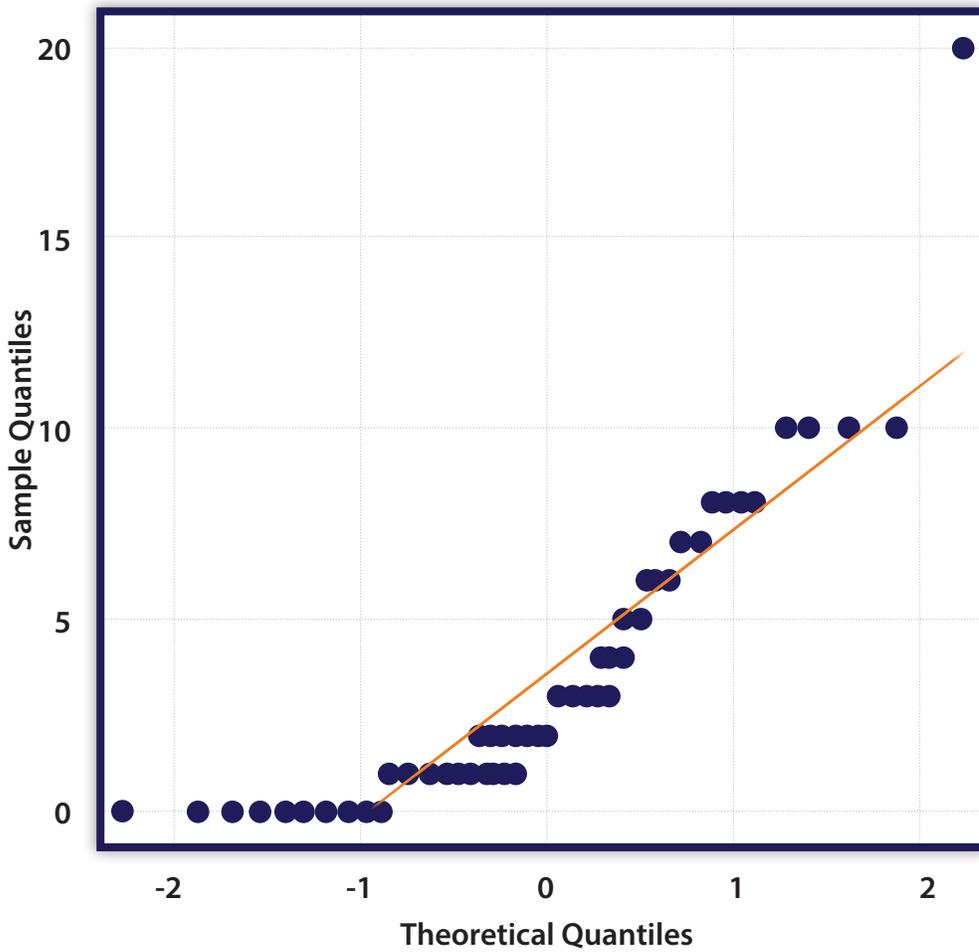
The mean number of surgeries is 2.9 with a range from 0 to 10.

Surgeries included removal of adnoids/tonsils, eye surgeries, feeding tube placement, dental surgeries, hernia repairs, ear tube placement, kidney/bladder surgery, GI surgeries, cardiac issues, and tongue tie/snip.

Negative numbers indicate the answer was left blank.



Normal Quantile Plot | Number of Hospitalizations

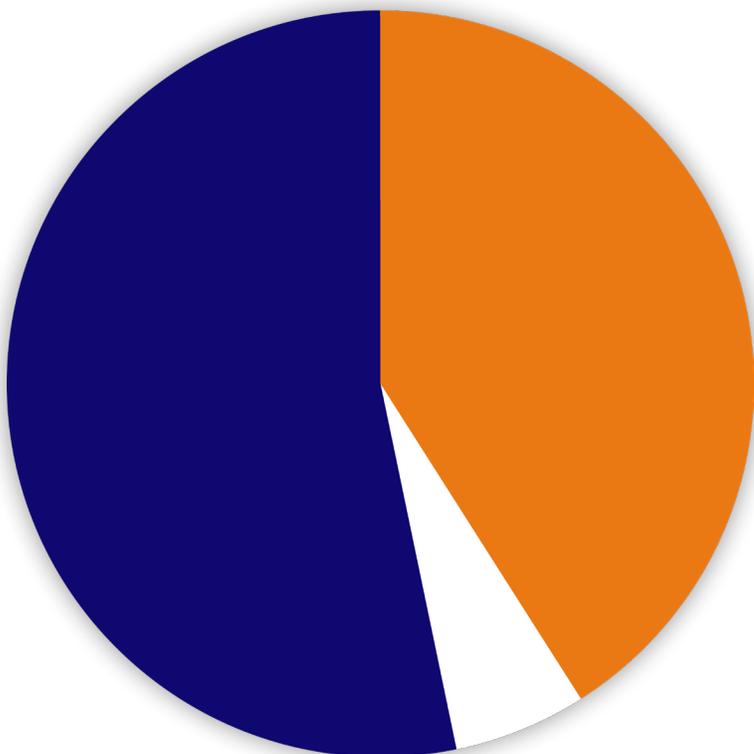


Notes

Mean Hospitalizations: 3.8 with a range from 0-10.

Negative numbers indicate the answer was left blank.

Cardiac Issues



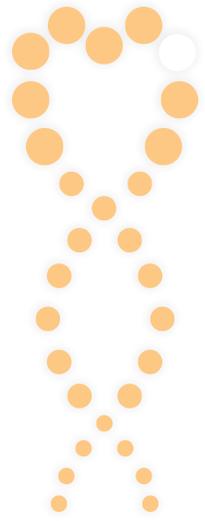
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- No
- Yes
- No Information

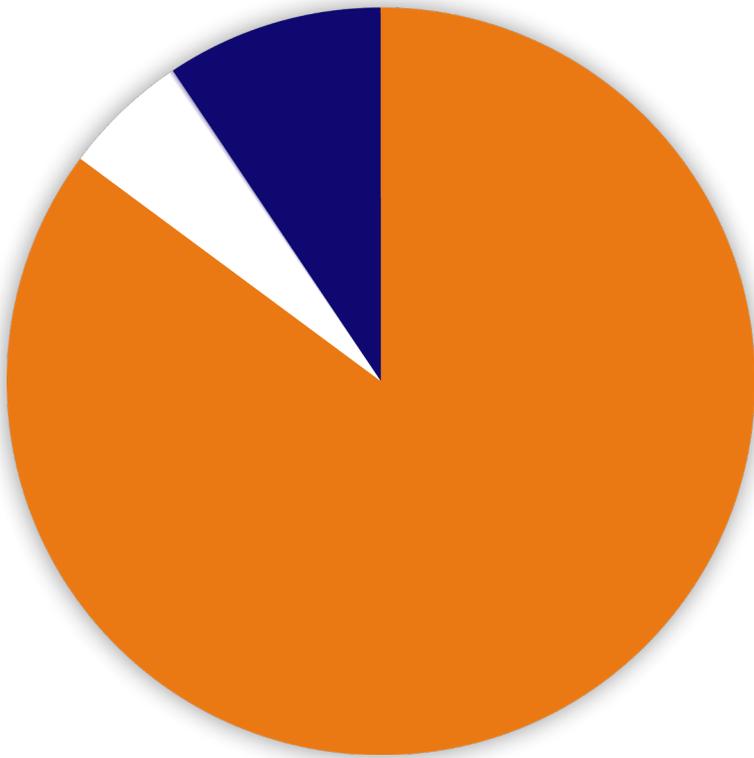
Notes

Of the polled respondents, 38% indicated issues related to the heart.

Symptoms listed were: Heart Murmur, Aortic Stenosis, Patent Ductus Arteriosus (PDA), Atrial Septal Defect (ASD), Ventricular Septal Defect (VSD), Tachychardia, and Patent Foramen Ovale (PFO).



Sleep Issues



Key

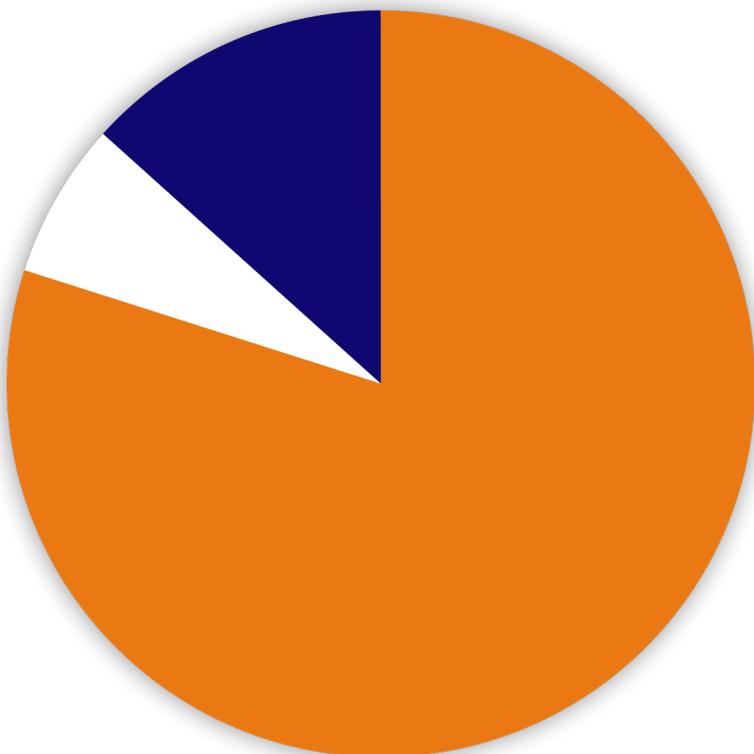
-  No
-  Yes
-  No Information

Notes

Of the polled respondents, 83% indicated difficulties related to sleep.

Symptoms include: sleep apnea, delayed sleep onset, frequent nighttime waking, and one instance of narcolepsy.

Eye Issues



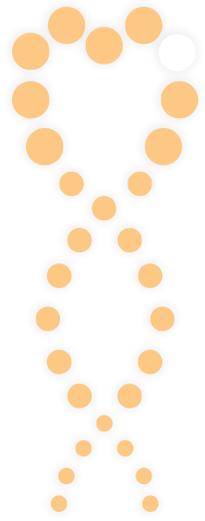
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-  No
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-  No Information

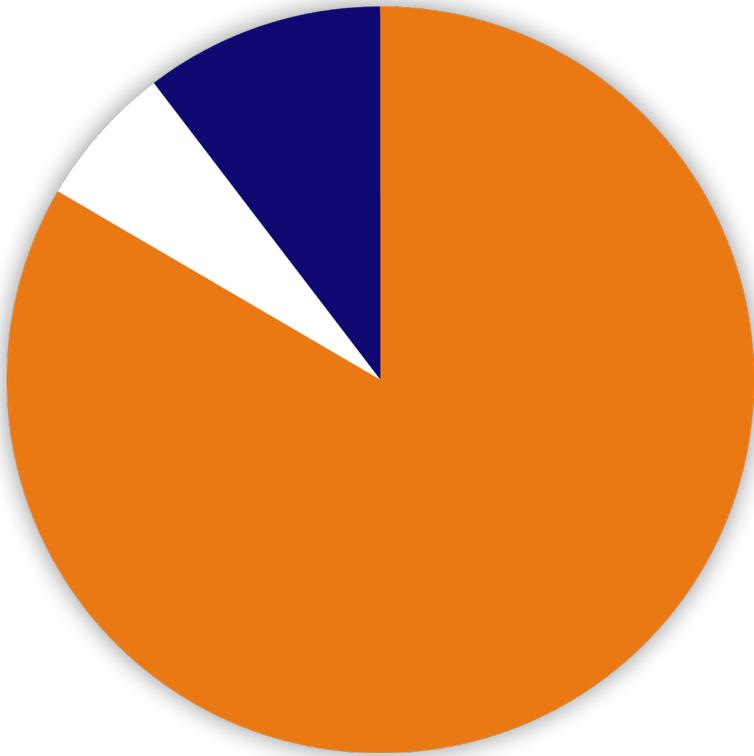
Notes

Of the polled respondents, 77% indicated difficulties related to the eyes.

Symptoms include: ptosis, strabismus, epicanthal folds, Hyperopia (far-sighted), Myopia (near-sighted).



Mouth | Palate | Dental Issues



Key

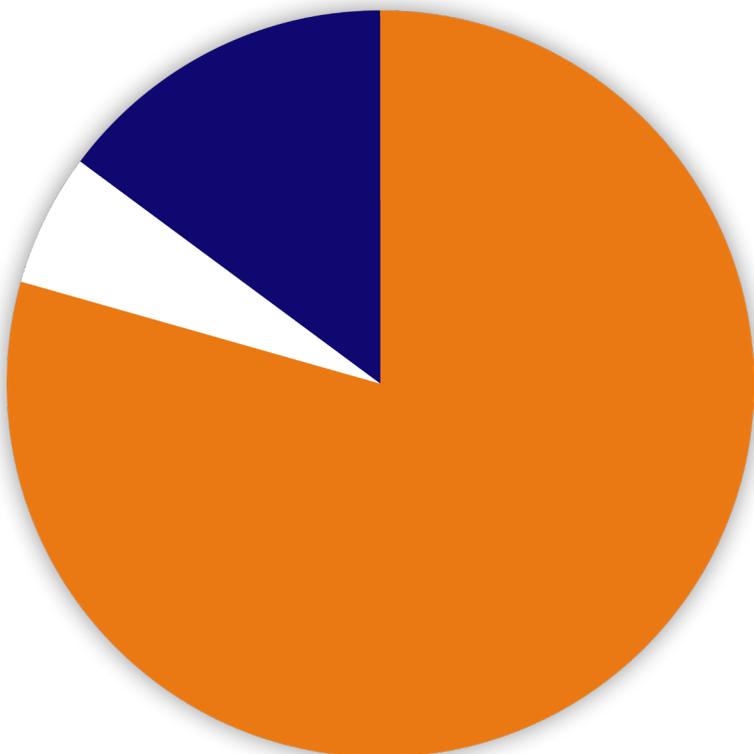
-  No
-  Yes
-  No Information

Notes

Of the polled respondents, 85% indicated issues related to the mouth.

Overwhelmingly, respondents indicated that early loss of primary teeth was the number one issue (a diagnostic feature of WSS). Followed by a high, arched palate and being tongue tied.

Hair | Skin Issues



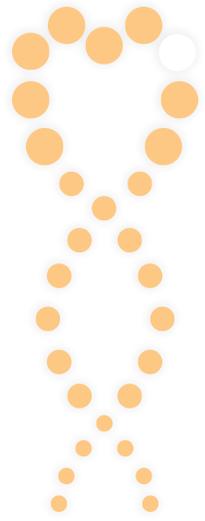
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-  No Information

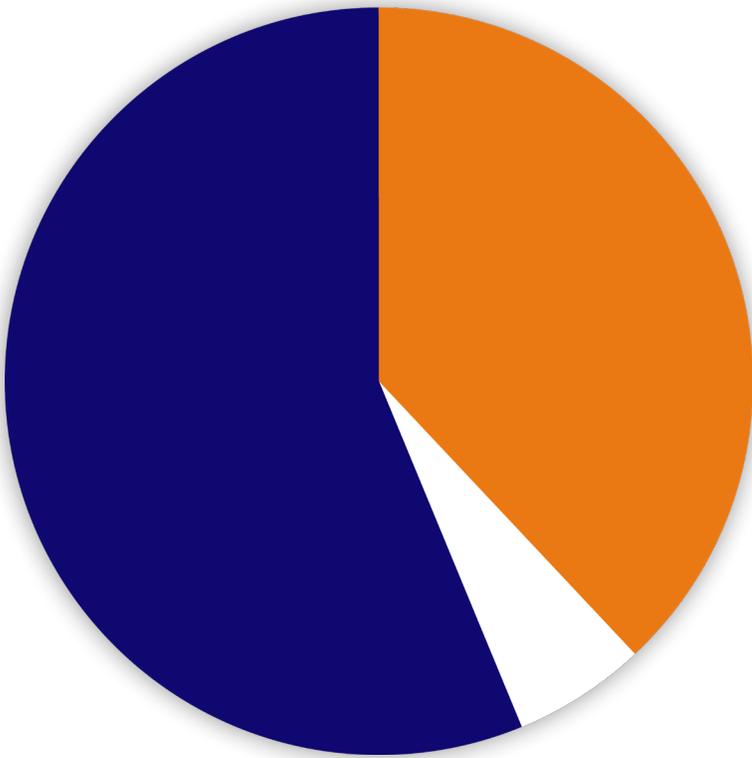
Notes

Of the polled respondents, 82% indicated hair and/or skin issues.

Hypertrichosis (abnormal hair XXXX) is a known, distinguishing feature of WSS. Followed by Eczema, Edema, and purplish/white extremities (Raynaud's Phenomenon).



Bladder | Kidney Issues



Key

-  No
-  Yes
-  No Information

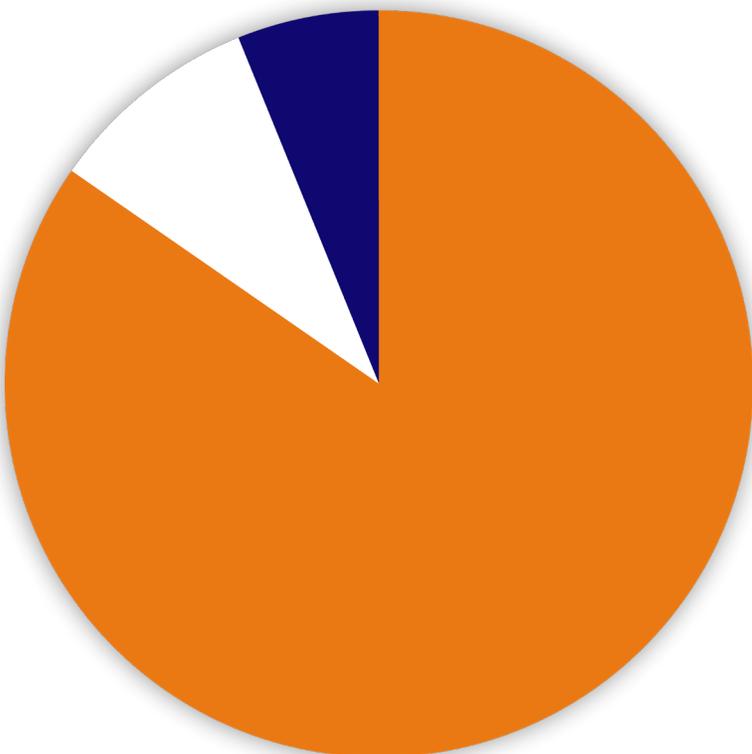
Notes

Of the polled respondents, 28% indicated bladder or kidney issues.

Symptoms include: Hydronephrosis (swelling of the kidney), neurogenic bladder, and vesicoureteral reflux.

Other symptoms include: rotated kidney, small cysts, duplicated collecting systems, and renal tubular acidosis.

Neurological Issues



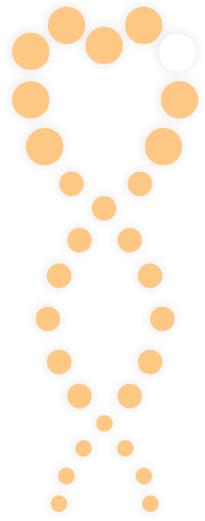
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-  No
-  Yes
-  No Information

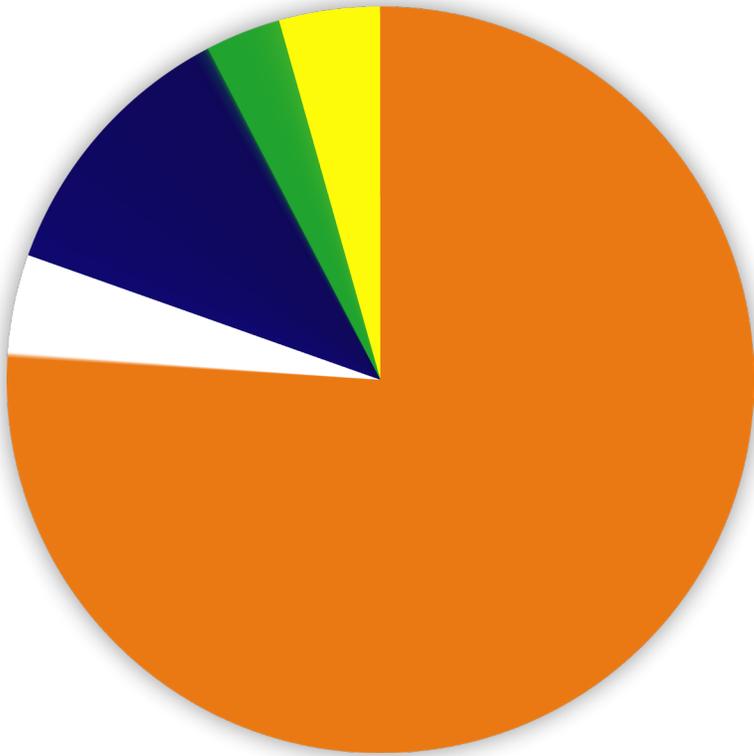
Notes

Of the polled respondents, 81% indicated neurological issues.

Issues described include: agenesis of the corpus callosum, aggressive behavior, ADHD, Autism Spectrum Disorder, Epilepsy, High Pain Threshold, and Hypotonia.



Primary Means of Feeding Intake



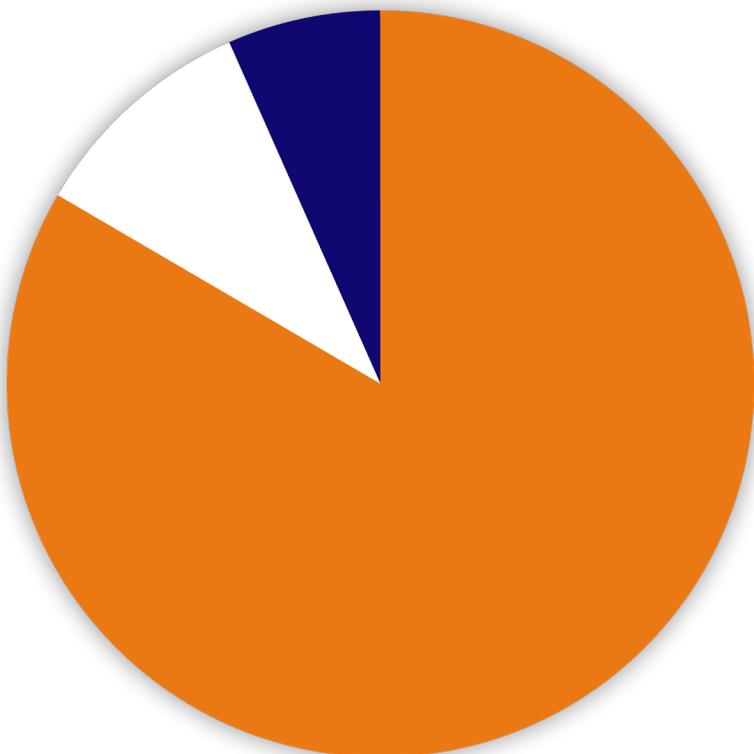
Key

- Oral
- Tube Feeding (GJ-t)
- Tube Feeding (G-tu)
- Tube Feeding (J-tu)
- No Information

Notes

Of the WSS patients polled, 77% are able to take food orally and 21% are tube-fed.

Difficulty with Digestion



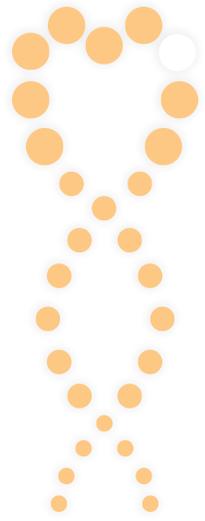
Key

- Yes
- No
- No Information

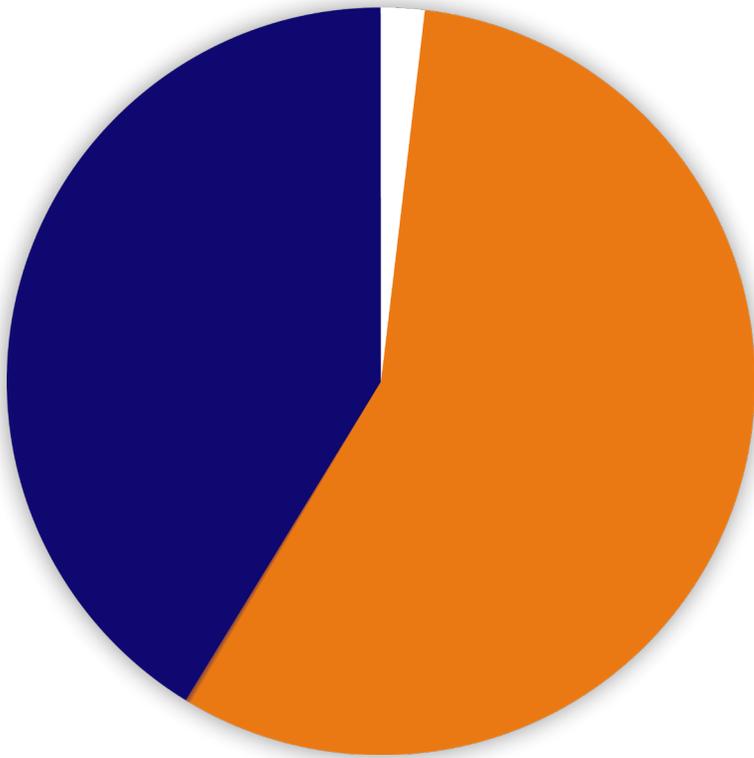
Notes

Of the polled respondents, 93% have digestion issues.

Symptoms include: Failure to thrive is the primary symptom, aspiration, constipation, delayed motility, chronic diarrhea.



NICU After Birth



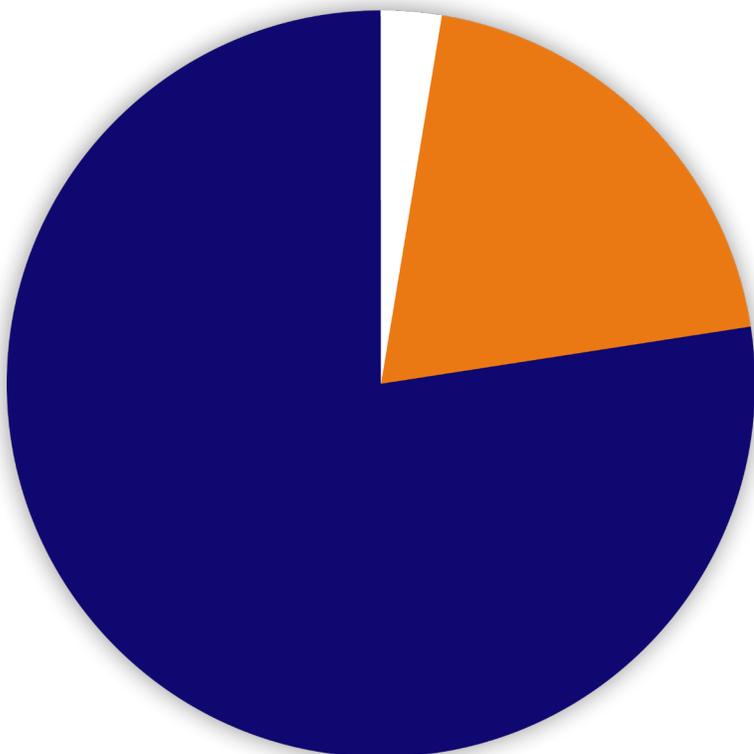
Key

- No
- Yes
- No Information

Notes

Of the polled respondents, 57% of WSS children spend time in the NICU after birth.

Other Identified Genetic Mutations



Key

- No
- Yes
- No Information

Notes

Of the polled respondents, 21% indicated they hold a second or third diagnosis of genetic disorders.