

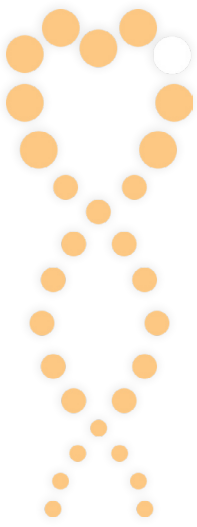


WSS Foundation

Wiedemann-Steiner Syndrome Foundation

CoRDS Registry Data

February 2019



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 web-page 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

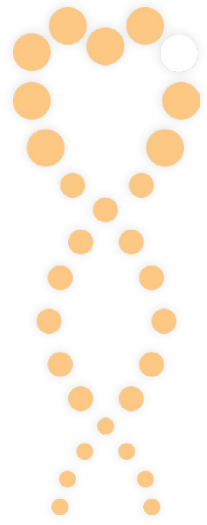
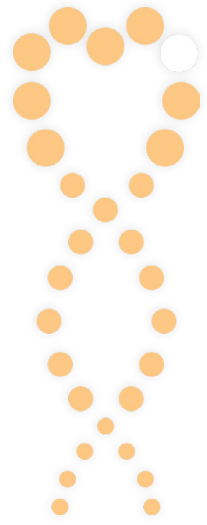


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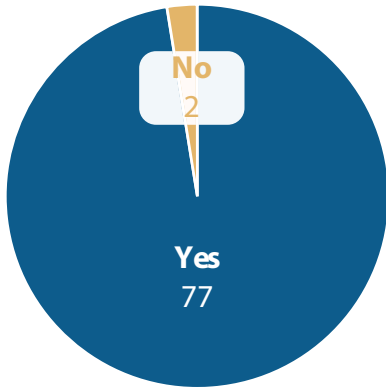
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Problems with Digestion

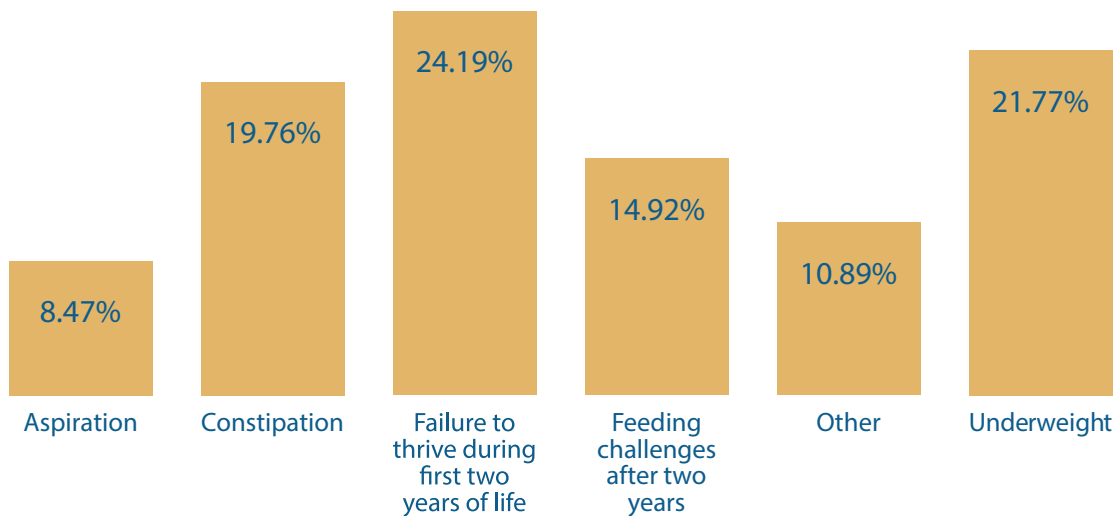
Has the participant ever had any problems with Digestion or feeding?

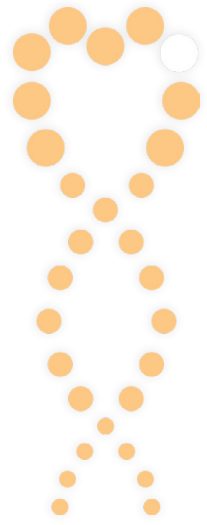
Has the participant ever had any problems with digestion or feeding?	Count	Percent
Yes	77	97.47%
No	2	2.53%



If yes, please select the symptoms that the participant has experienced.

Symptoms	Count	Percent
Aspiration	21	8.47%
Constipation	49	19.76%
Failure to thrive during first two years of life	60	24.19%
Feeding challenges after two years	37	14.92%
Other	27	10.89%
Underweight	54	21.77%

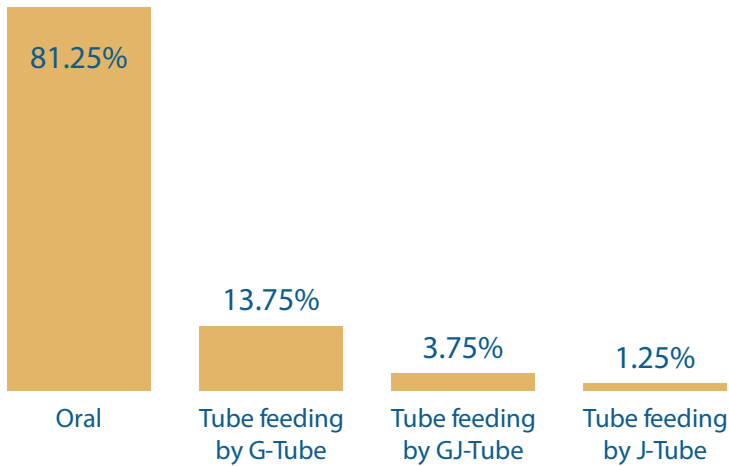




Problems with Digestion (Cont.)

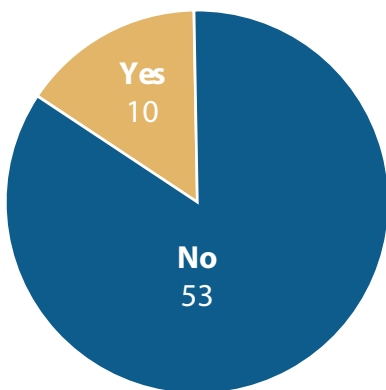
What's the participant's primary means of intake?

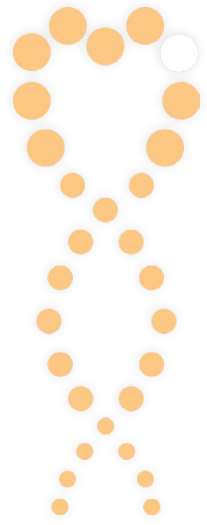
Primary means of intake	Count	Percent
Oral	65	81.25%
Tube feeding by G-tube	11	13.75%
Tube feeding by GJ-tube	3	3.75%
Tube feeding by J-tube	1	1.25%



If orally fed, has the participant ever previously required tube feeding?

If orally fed, has the participant ever previously required tube feeding?	Count	Percent
No	53	84.13%
Yes	10	15.87%

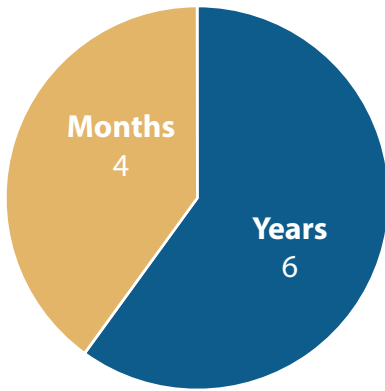




Problems with Digestion (Cont.)

If yes, at what age did the participant begin weaning off of tube feeding?

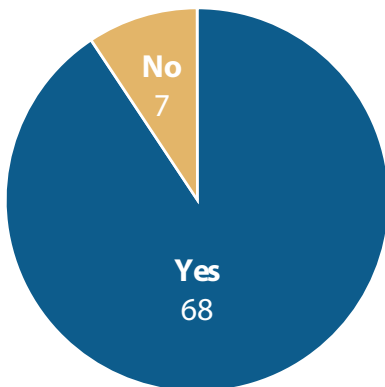
Age	Count	Percent
Months	4	40%
Years	6	60%

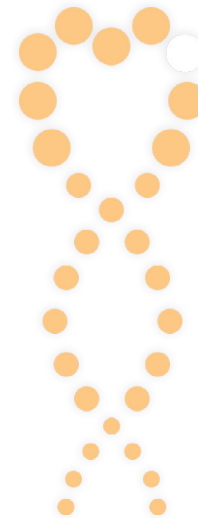


Neurological Issues

Has the participant ever been diagnosed with any neurological issues?

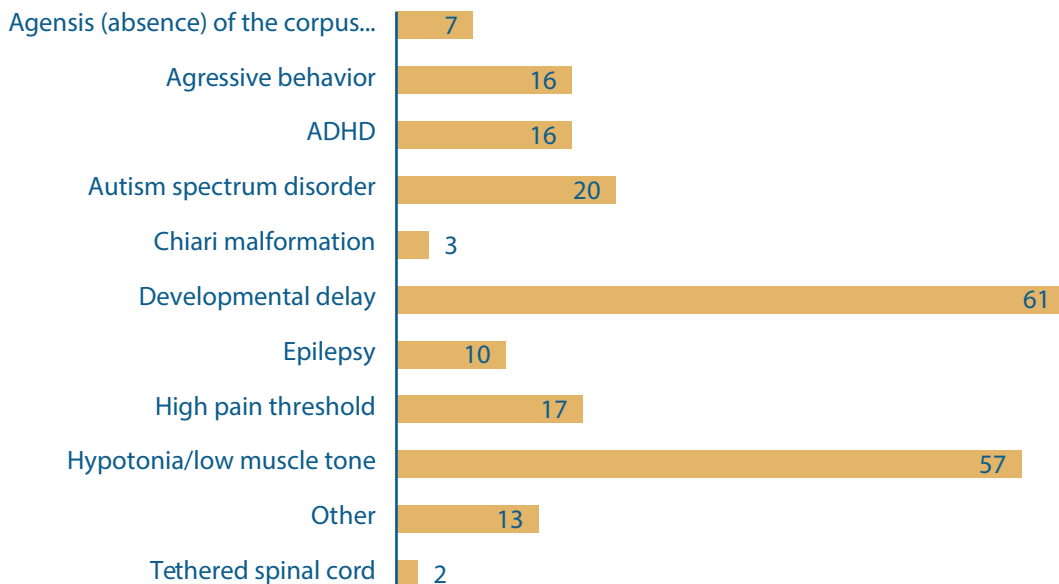
Has the participant ever been diagnosed with any Neurological issues?	Count	Percent
Yes	68	90.67%
No	7	9.33%

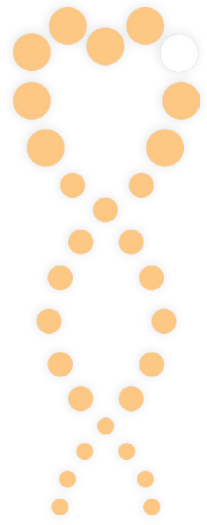




Has the participant ever been diagnosed with any neurological issues?

Has the participant ever been diagnosed with any Neurological issues?	Count	Percent
Agenesis (absence) of the corpus callosum of the brain	7	3.15%
Aggressive behavior	16	7.21%
Attention deficit hyperactivity disorder (ADHD)	16	7.21%
Autism spectrum disorder	20	9.01%
Chiari malformation	3	1.35%
Developmental delay/intellectual disability	61	27.48%
Epilepsy	10	4.5%
High pain threshold	17	7.66%
Hypotonia/low muscle tone	57	25.68%
Other	13	5.86%
Tethered spinal cord	2	.9%

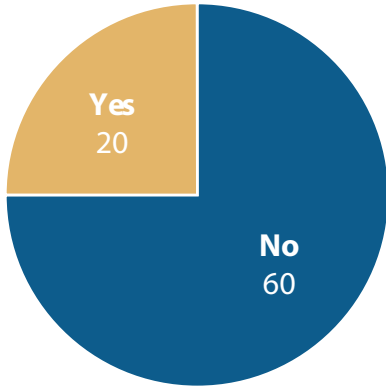




Bladder or Kidney Issues

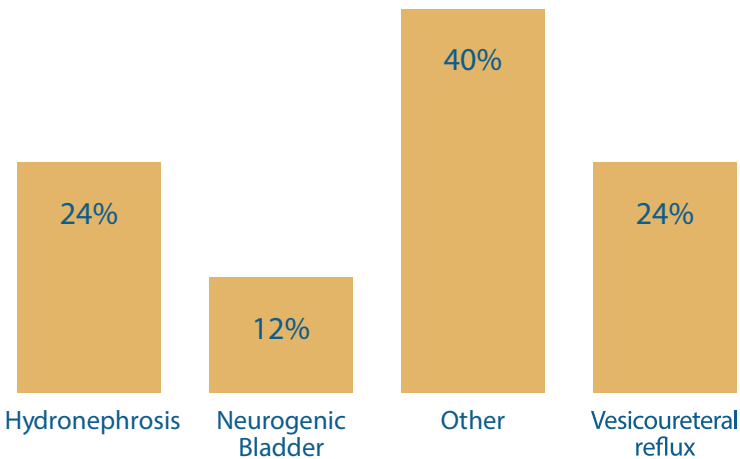
Had the participant ever had any bladder or kidney issues?

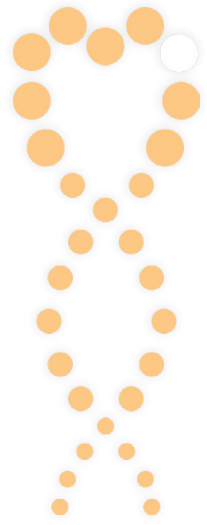
Had the participant ever had any bladder or kidney issues?	Count	Percent
No	60	75%
Yes	20	25%



If yes, please select the symptoms that the participant has experienced.

Symptoms	Count	Percent
Hydronephrosis (kidney swelling)	6	24%
Neurogenic bladder	3	12%
Other	10	40%
Vesicoureteral reflux	6	24%

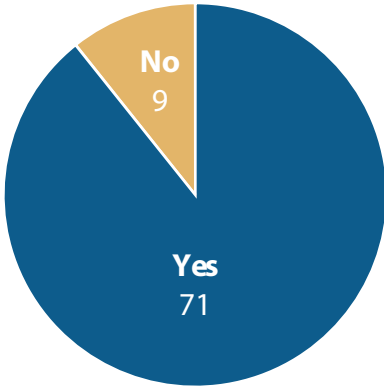




Mouth, Palate, or Dental Issues

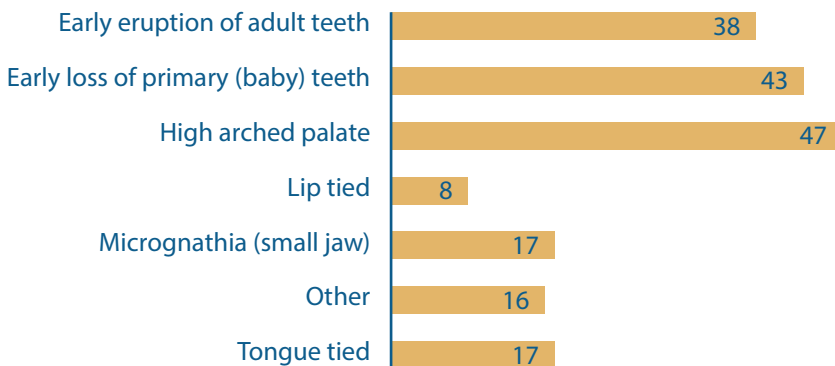
Has the participant ever had any mouth, palate, or dental issues?

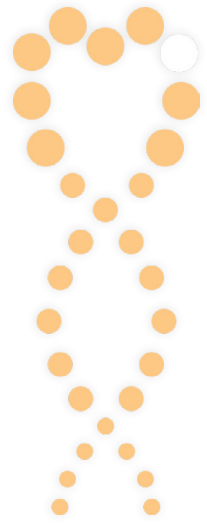
Has the participant ever had any mouth, palate or dental issues?	Count	Percent
No	9	11.25%
Yes	71	88.75%



If yes, please select the symptoms that the participant has experienced.

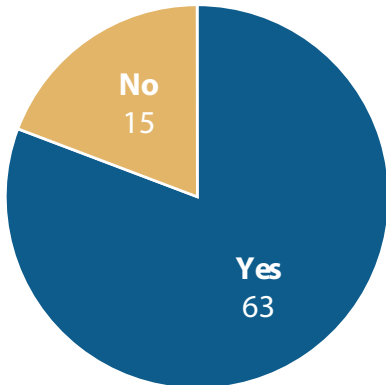
Symptoms	Count	Percent
Early eruption of adult teeth	38	20.43%
Early loss of primary (baby teeth)	43	23.12%
High arched palate	47	25.27%
Lip tied	8	4.3%
Micrognathia (small jaw)	17	9.14%
Other	16	8.6%





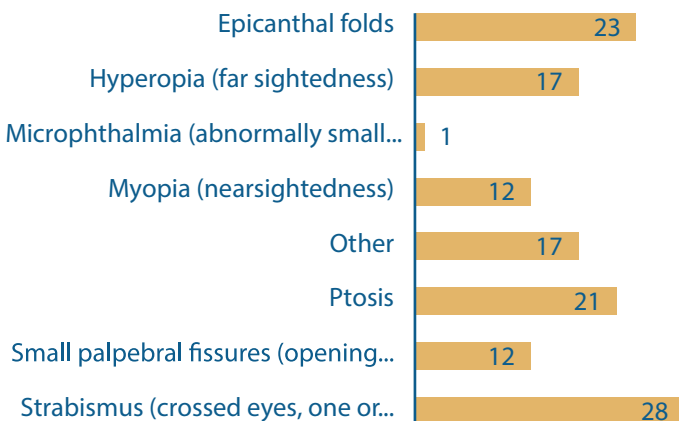
Has the participant ever had any eye or vision problems?

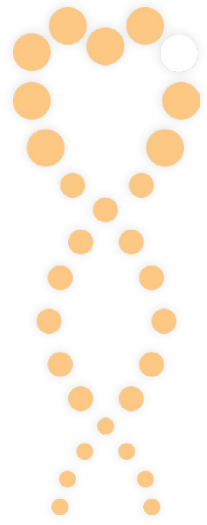
Has the participant ever had any eye or vision problems?	Count	Percent
No	15	19.23%
Yes	63	80.77%



If yes, please select the symptoms that the participant has experienced.

Symptoms	Count	Percent
Epicanthal folds	23	17.56%
Hyperopia (far sightedness)	17	12.98%
Microphthalmia (abnormally small eyeball)	1	.76%
Myopia (nearsightedness)	12	9.16%
Other	17	12.98%
Ptosis (drooping of upper eyelid)	21	16.03%
Small palpebral fissures (opening between eyelids)	12	9.16%
Strabismus (crossed eyes, one or both eyes turn inward or outward)	28	21.37%

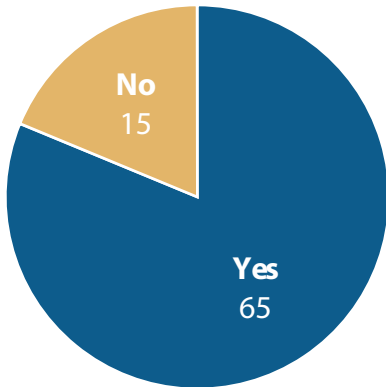




Skeletal Issues

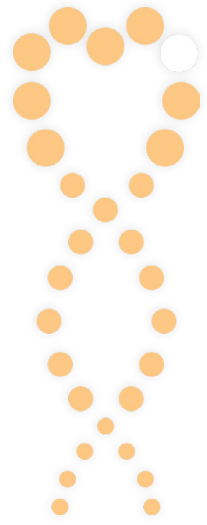
Has the participant ever had skeletal (bone) issues?

Has the participant ever had skeletal (bone) issues?	Count	Percent
No	15	18.75%
Yes	65	81.25%



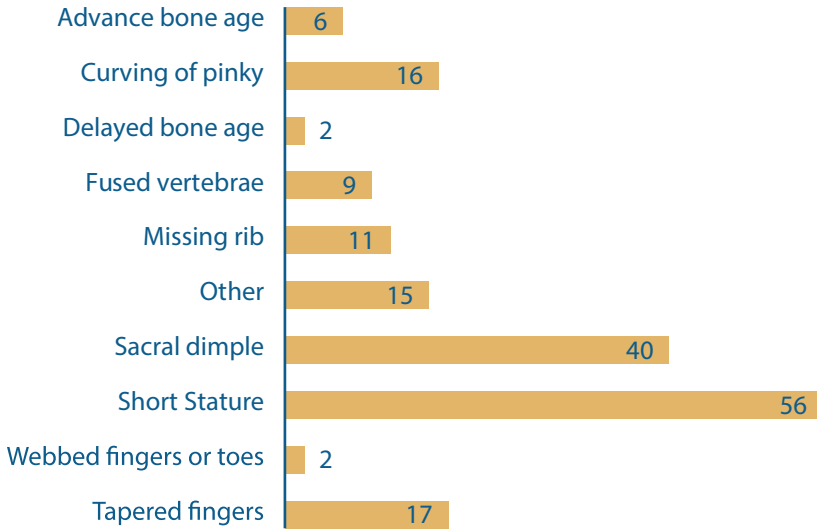
If yes, please select the symptoms that the participant has experienced.

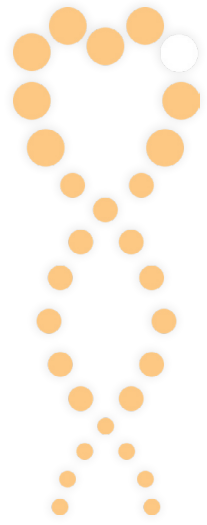
Symptoms	Count	Percent
Advanced bone age	6	3.45%
Clinodactyly of fifth finger (curving of pinky)	16	9.2%
Delayed bone age	2	1.15%
Fused vertebrae	9	5.17%
Missing rib	11	6.32%
Other	15	8.62%
Sacral dimple	40	22.99%
Short Stature	56	32.18%
Syndactyly (two or more digits fused together, webbed fingers or toes)	2	1.15%
Tapered finger(s)	17	9.77%



Skeletal Issues (Cont.)

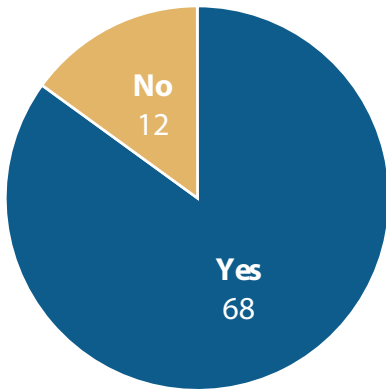
If yes, please select the symptoms that the participant has experienced. (Cont.)





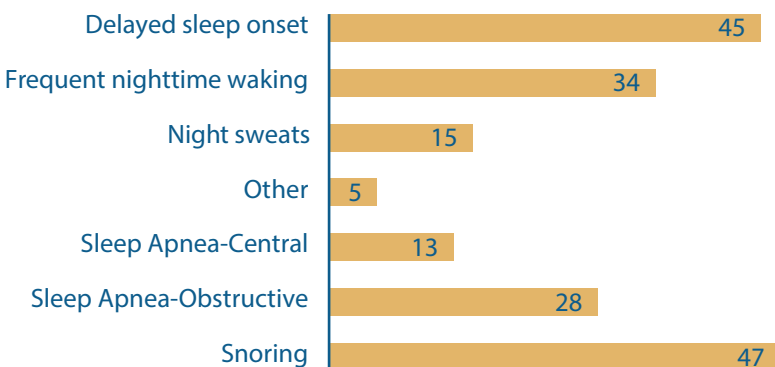
Has the participant ever had sleep issues?

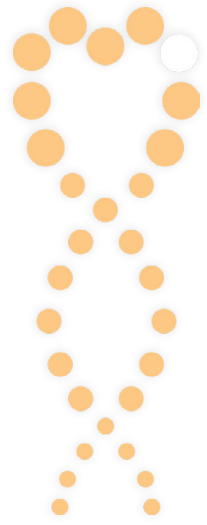
Has the participant ever had sleep issues?	Count	Percent
No	12	15%
Yes	68	85%



If yes, please select the symptoms that the participant has experienced.

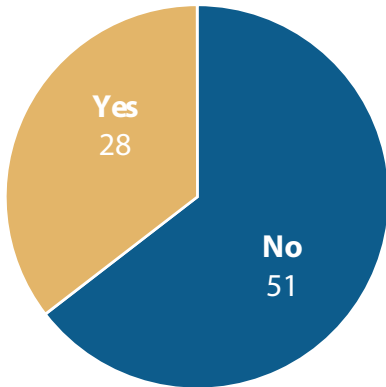
Symptoms	Count	Percent
Delayed sleep onset (difficulty falling asleep)	45	24.06%
Frequent nighttime waking	34	18.18%
Night Sweats	15	8.02%
Other	5	2.67%
Sleep Apnea-Central (brain's inability to tell muscles to breath)	13	6.95%
Sleep Apnea-Obstructive (intermittent air flow blockage during sleep)	28	14.97%
Snoring	47	25.13%





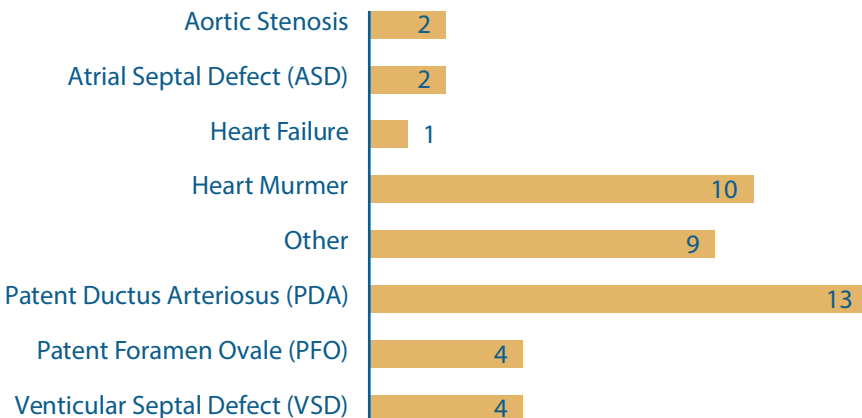
Has the participant ever had any cardiac issues?

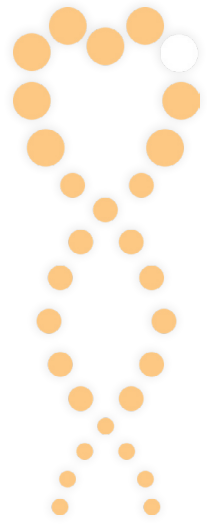
Has the participant ever had any cardiac issues?	Count	Percent
No	51	64.56%
Yes	28	35.44%



If yes, please select the symptoms that the participant has experienced.

Symptoms	Count	Percent
Aortic Stenosis (narrowing of aortic valve)	2	4.44%
Atrial Septal Defect (ASD) (hole between upper chambers of heart)	2	4.44%
Heart Failure	1	2.22%
Heart Murmur (unusual sound between heartbeats)	10	22.22%
Other	9	20.00%
Patent Ductus Arteriosus (PDA) (persistent opening between two major arteries)	13	28.89%
Patent Foramen Ovale (PFO) (hole in the heart that does not close)	4	8.89%
Ventricular Septal Defect (VSD) (hole between lower chambers of heart)	4	8.89%

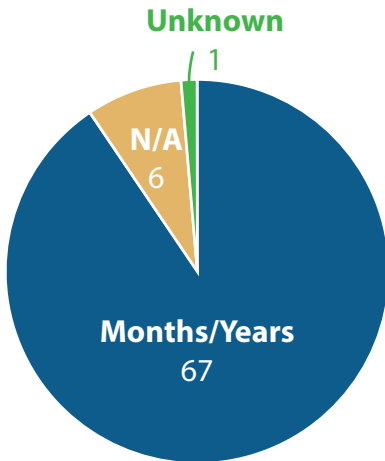




Development by Age

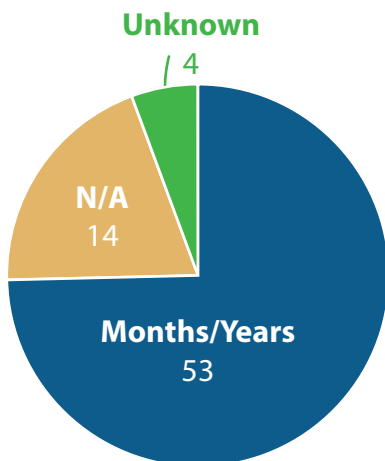
At what age was the participant able to walk?

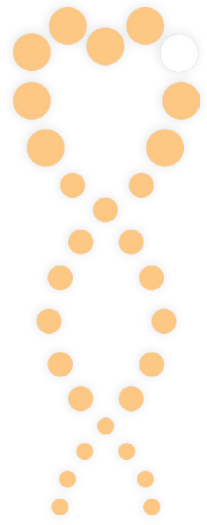
Age	Count	Percent
Months/Years	67	90.54%
Not applicable	6	8.11%
Unknown	1	1.35%



At what age was the participant able to speak?

Age	Count	Percent
Months/Years	53	74.65%
Not applicable	14	19.72%
Unknown	4	5.63%

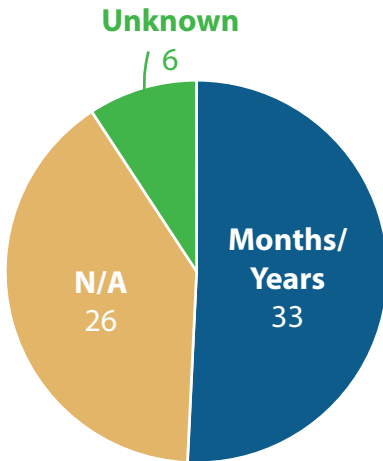




Development by Age (Cont.)

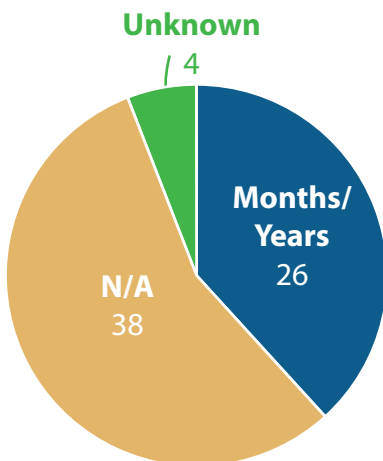
At what age was the participant able to speak phrases?

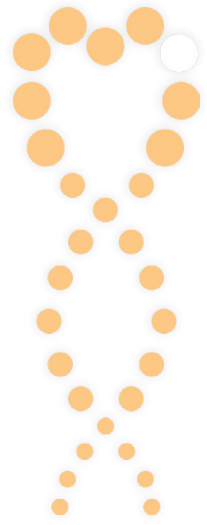
Age	Count	Percent
Months/Years	33	50.77%
Not applicable	26	40%
Unknown	6	9.23%



At what age did the participant achieve urinary continence?

Age	Count	Percent
Months/Years	26	38.24%
Not applicable	38	55.88%
Unknown	4	5.88%

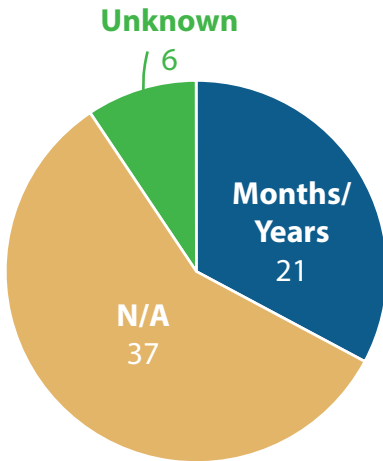




Development by Age (Cont.)

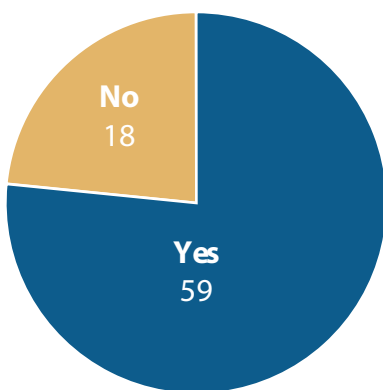
At what age did the participant achieve bowel continence?

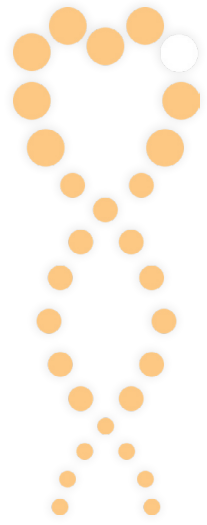
Age	Count	Percent
Months/Years	21	32.81%
Not applicable	37	57.81%
Unknown	6	9.38%



Has the participant or is the participant currently attending school?

Age	Count	Percent
No	18	23.38%
Yes	59	76.62%

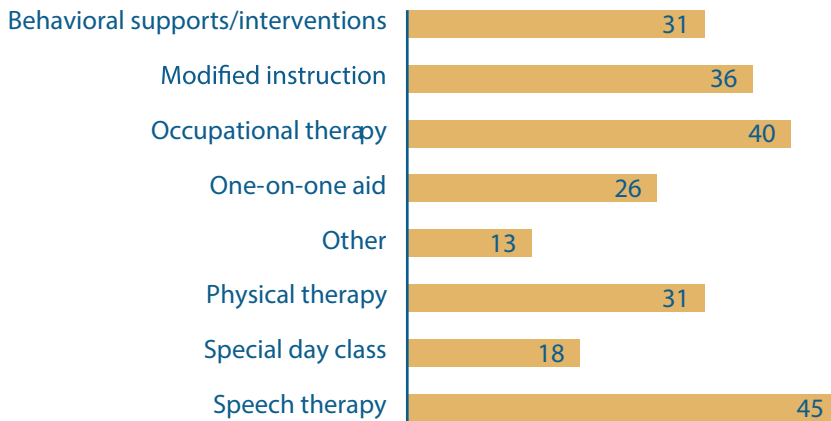


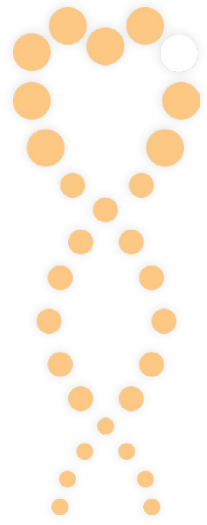


Development by Age (Cont.)

What type of school services/supports has the participant received upon entering kindergarten and beyond?

Services/Supports	Count	Percent
Behavioral supports/interventions	31	12.92%
Modified instruction	36	15%
Occupational therapy	40	16.67%
One-on-one aid	26	10.83%
Other	13	5.42%
Physical therapy	31	12.92%
Special day class	18	7.5%
Speech therapy	45	18.75%

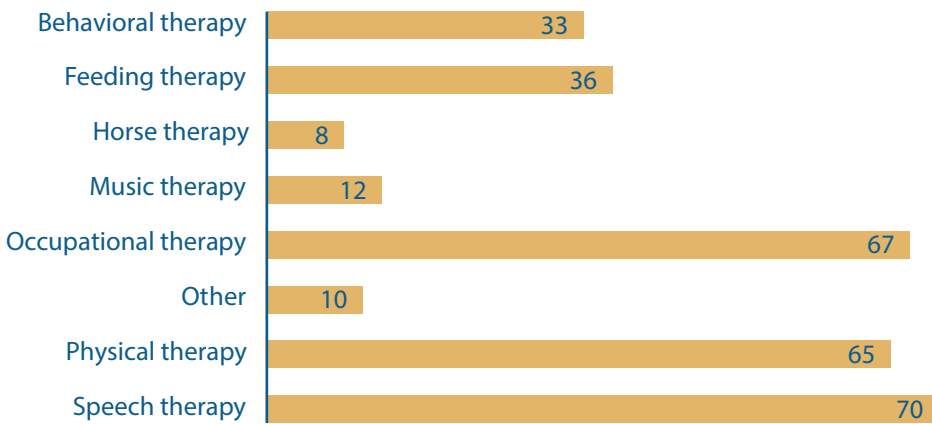




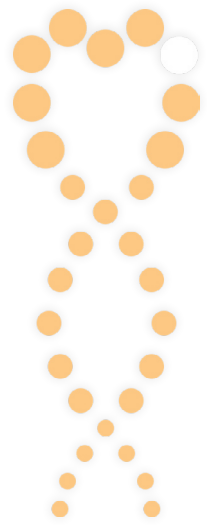
Development by Age (Cont.)

What type of services/therapy has the participant received?

Services/Supports	Count	Percent
Behavioral therapy	33	10.96%
Feeding therapy	36	11.96%
Horse therapy	8	2.66%
Music therapy	12	3.99%
Occupational therapy	67	22.26%
Other	10	3.32%
Physical therapy	65	21.59%
Speech therapy	70	23.26%



Development by Age (Cont.)



Variable (Years)	N	N Miss	Mean	Std Dev	Minimum	Maximum
Weaned off tube feeding	8	74	3.6	3.08	.25	10
Speak single words	54	28	2.01	1.23	.5	6.25
Walk	66	16	1.96	.93	.83	6.17
Speak phrases	43	39	2.97	1.24	.75	6.25
Bladder trained	33	49	4.05	2.17	.33	12
Bowel trained	30	52	4.83	3.19	.33	16
IQ Score	14	68	66.57	14.19	40	85