

NCBRS Patient Registry Data

In collaboration with CoRDS

May 2021

SANFORD
RESEARCH

CoRDS



NCBRS
WORLDWIDE
FOUNDATION



Intro

Welcome to the first edition of the Patient Registry data report.

Do you remember that feeling you had when you first received the diagnosis of Nicolaides Baraitser Syndrome (NCBRS). Going online to search for information, statistics, data and finding nothing available to give you more insights into the condition. That's what I found when my son Callum was diagnosed in 2007.

This inspired my wife Michelle - NCBRS Board Member and I to start a UK support group in 2010, holding a meeting with the geneticists and five other families attending from around the world. In 2015 we first meet Helen and her son Kyle at a UK meeting, this is when we realised the need for a website which has been an invaluable source of information for not only patients and their families but for doctors and professionals alike. Fast forward to 2020 and this is when Helen, NCBRS Co-Founder/Chair and I co-founded the NCBRS Worldwide Foundation and partnered with CoRDS - Coordination of Rare Diseases at Sanford to gather data and insights to better understand NCBRS, best treatments and to contribute to any future research studies .

Lee Reavey - NCBRS Co-Founder/CEO

What is CoRDS?

CoRDS is a centralised international registry that stores detailed information about individuals who are affected by a specific condition. For rare syndromes such as NCBRS, they are essential in connecting patients and researchers in order to advance knowledge and treatments. Having a registry will allow us to develop an official worldwide number of documented cases of NCBRS. The information provided is stored securely and no unauthorised people is able to gain access to any information about you. All information is de-identified.

As researchers and patients with NCBRS are located all around the world, having a centralised database makes it easier for clinical information to be shared. The register provides an excellent opportunity for families to be involved in research studies and clinical trials. Once questionnaires are completed researchers can compare specific symptoms, identify how the syndrome progresses as well as gain valuable insights into which treatments were most successful. Participation in the registry is completely voluntary.

In this report you will see a break down of the latest data available from the de-identified data submitted from our NCBRS participants. We have 53 participants enrolled with 40 sharing their data with us.

Helen Robinson - NCBRS Co-Founder/Chair



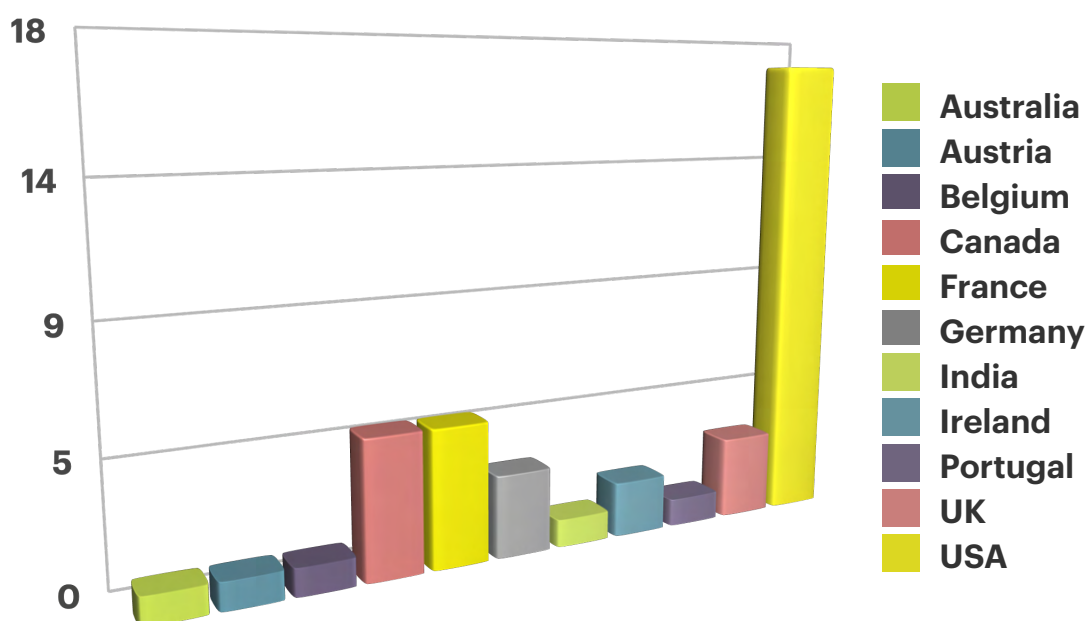
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General data

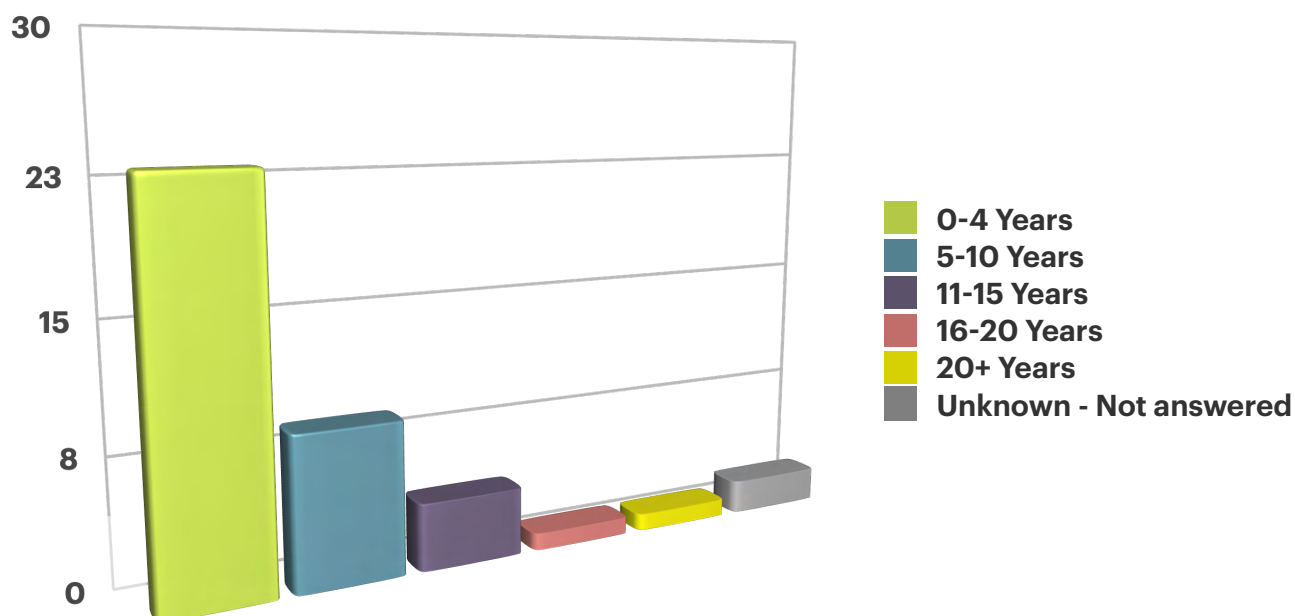
Participants and countries represented in this report.

Country	Count	Percentage
Australia	1	2.5%
Austria	1	2.5%
Belgium	1	2.5%
Canada	5	12.5%
France	5	12.5%
Germany	3	7.5%
India	1	2.5%
Ireland	2	5%
Portugal	1	2.5%
United Kingdom (UK)	3	7.5%
United States of America (USA)	17	42.5%
Totals	40	100%



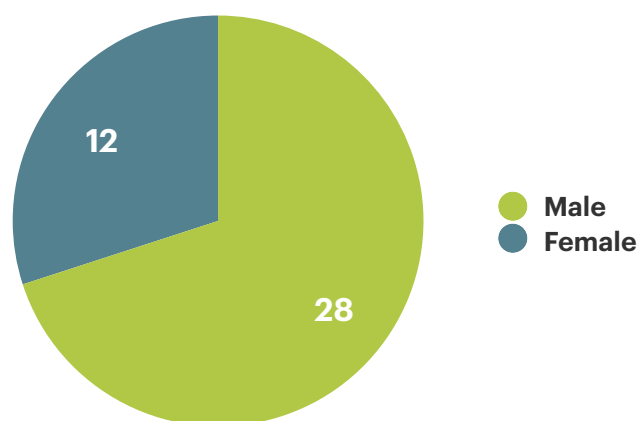
Age participant was diagnosed with NCBRS.

Age Diagnosed	Count	Percentage
0-4 Years	23	57.75%
5-10 Years	9	22.5%
11-15 Years	4	10%
16-20 Years	1	2.5%
20+ Years	1	2.5%
Unknown - Not answered	2	5%



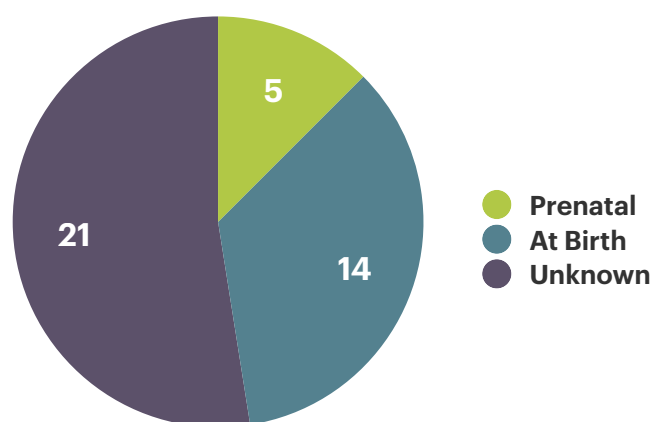
Participants Sex

Sex	Count	Percentage
Male	28	70%
Female	12	30%



Age symptoms where first recognised leading to a diagnosis of NCBRS?

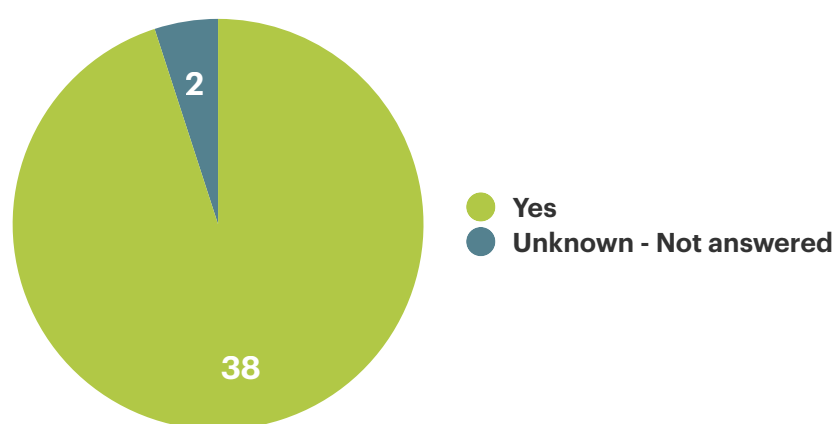
Symptoms First Recognised	Count	Percentage
Prenatal	5	12.5%
At Birth	14	35%
Unknown / Not answered	21	52.5%



Diagnosis

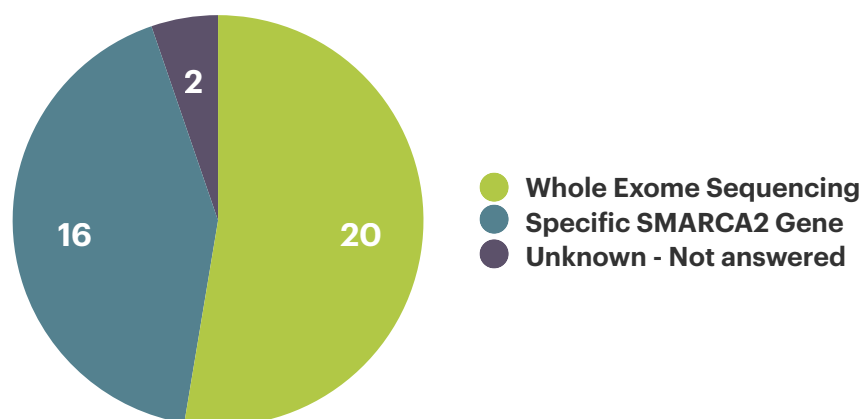
Was the participants diagnosis determined by genetic testing?

Diagnosed by genetic testing	Count	Percentage
Yes	38	95%
Unknown - Not answered	2	5%



If "yes" what type of genetic testing was utilised?

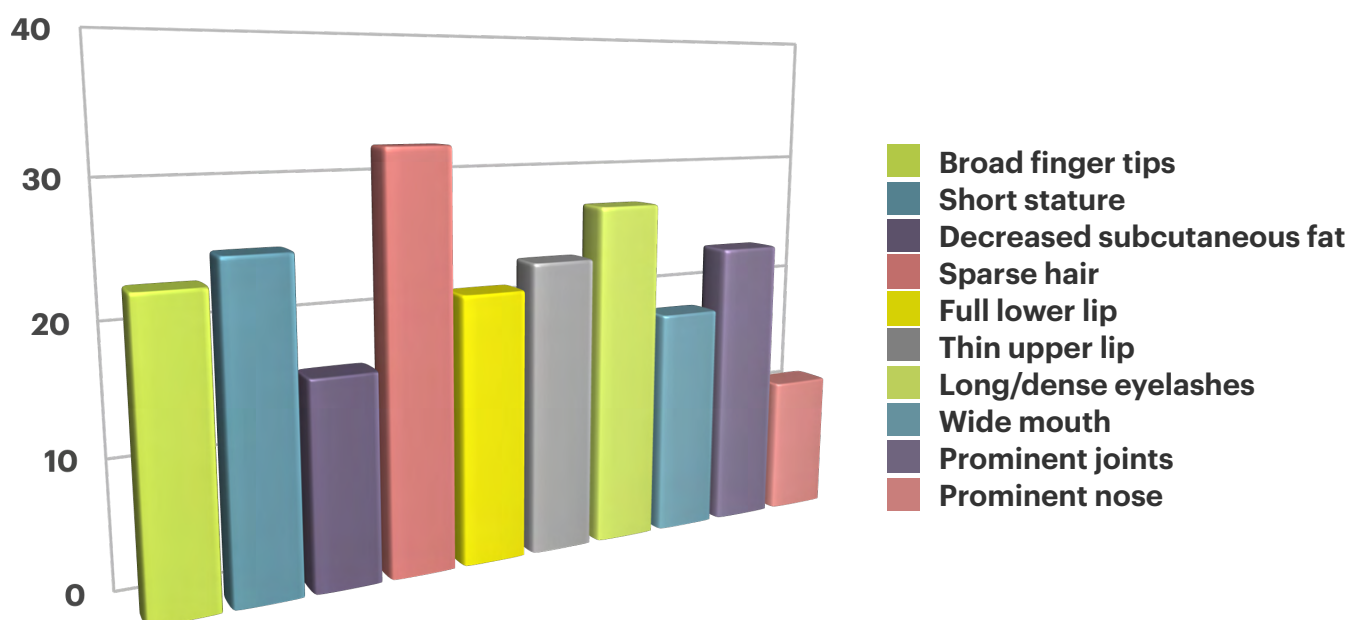
Test Utilised	Count	Percentage
Whole exome sequencing	20	52.63%
Specific SMARCA2 gene	16	42.10%
Unknown - Not answered	2	5.27%



Physical Characteristics

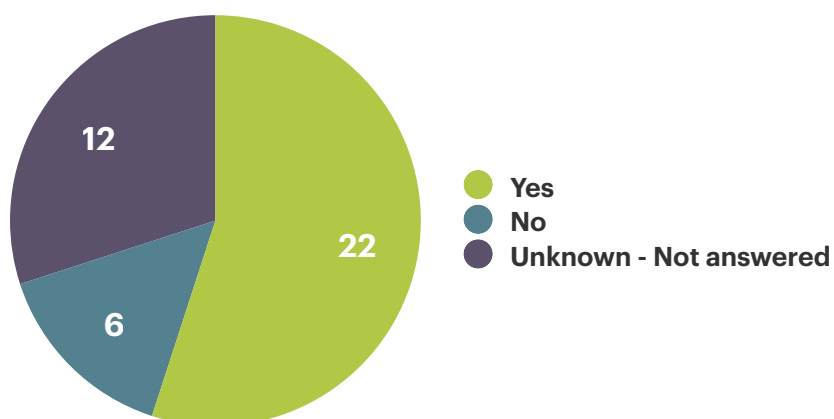
Which of the following unique features does the participant have?

Unique features	Count	Percentage
Broad finger tips	23	57.5%
Short stature	25	62.75%
Decreased subcutaneous fat in the cheeks	16	40%
Sparse hair	32	80%
Full lower lip	21	52.5%
Thin upper lip	23	57.5%
Long/dense eyelashes	27	67.5%
Wide mouth	18	45%
Prominent joints	23	57.5%
Prominent nose	11	27.5%



Has the unique features become more prominent as participant has aged?

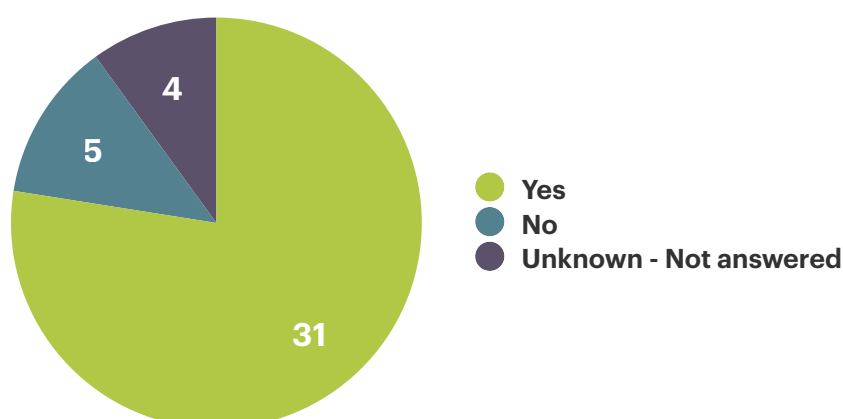
Features more prominent	Count	Percentage
Yes	22	55%
No	6	15%
Unknown - Not answered	12	30%



Symtoms

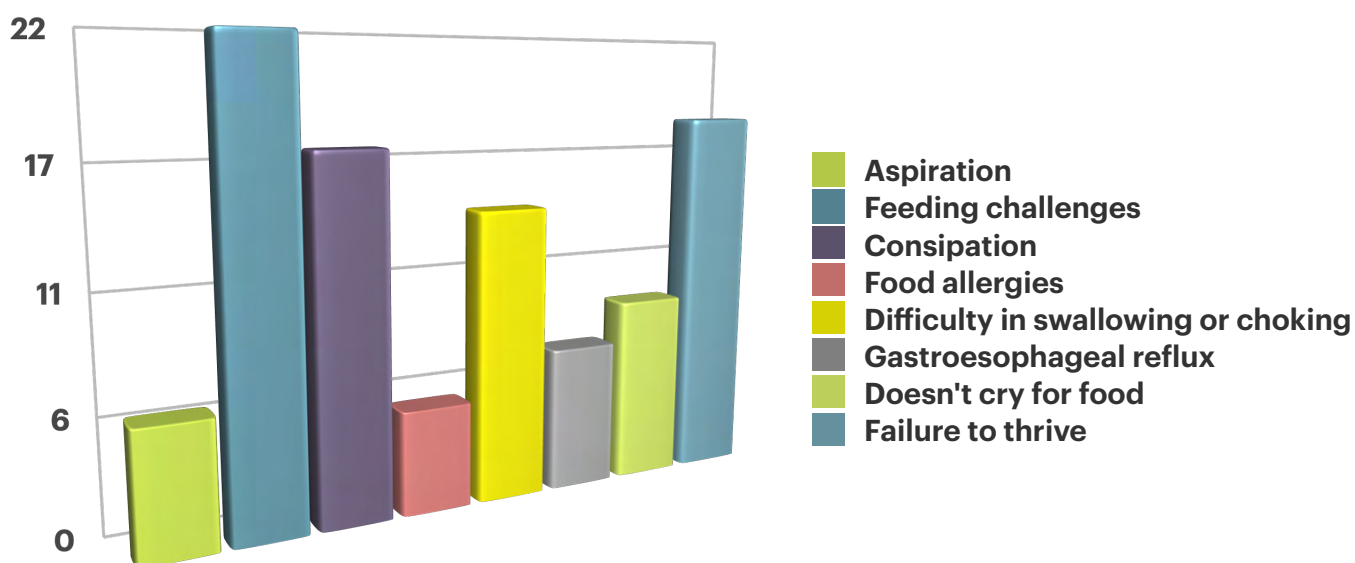
Has the participant ever had any problems with digestion or feeding?

Digestion or feeding problems	Count	Percentage
Yes	31	77.5%
No	5	12.5%
Unknown - Not answered	4	10%



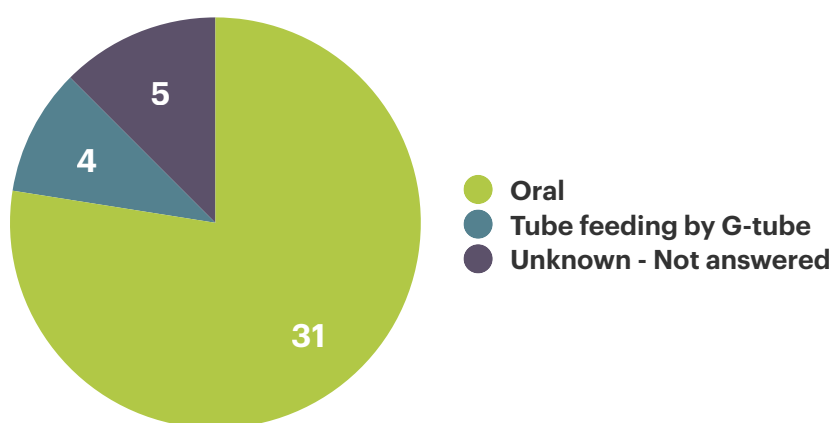
If “yes” please select the symptoms the participant has experienced.

Digestion or feeding problem	Count	Percentage
Aspiration	6	19.35%
Feeding challenges	22	70.96%
Constipation	17	54.83%
Food allergies	5	16.12%
Difficulty in swallowing or choking	14	45.16%
Gastroesophageal reflux	7	22.58%
Doesn't cry for food	9	29.03%
Failure to thrive	18	58.06%



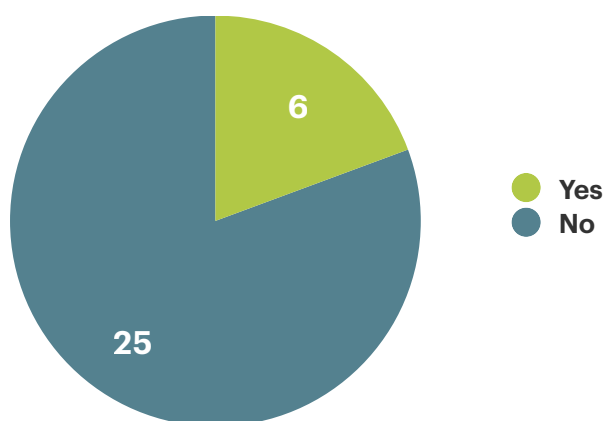
What is the participants primary means of intake?

Primary intake	Count	Percentage
Oral	31	77.5%
Tube feeding by G-tube	4	10%
Unknown / Not answered	5	12.5%



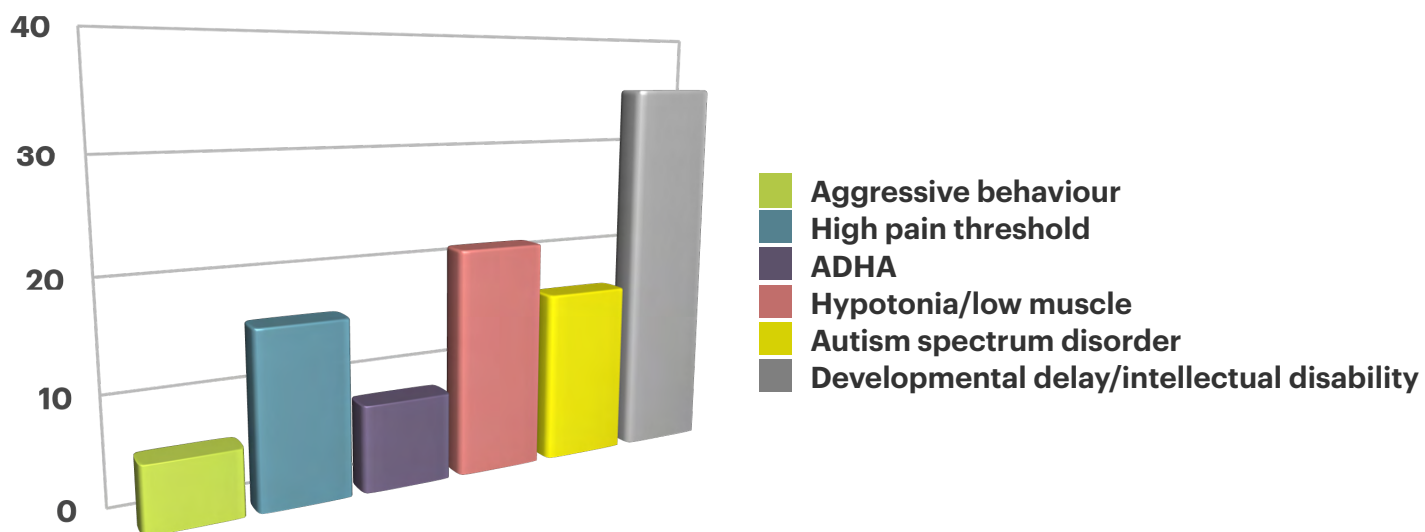
If orally fed, has the participant ever required tube feeding?

Tube feeding	Count	Percentage
Yes	6	19%
No	25	81%



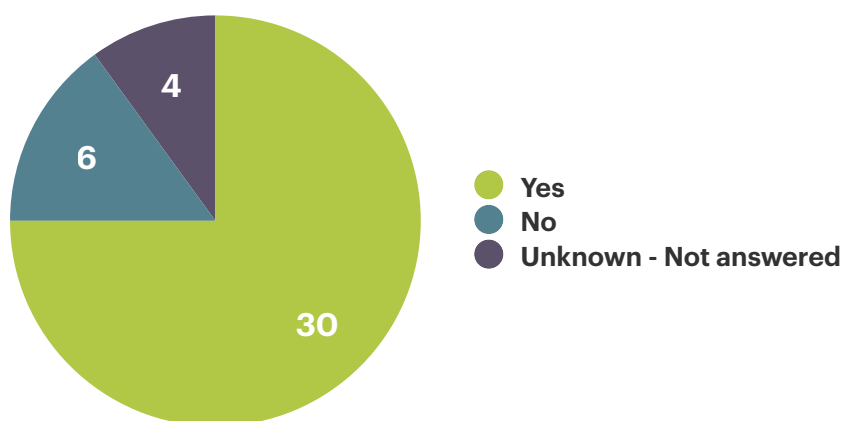
Has the participant ever been diagnosed with neurological issues?

Neurological issue	Count	Percentage
Aggressive behaviour	6	15%
High pain threshold	16	40%
Attention deficit, Hyperactive disorder (ADHD)	8	20%
Hypotonia/low muscle	21	52.5%
Autism spectrum disorder	16	40%
Development delay/intellectual disability	35	87.5%



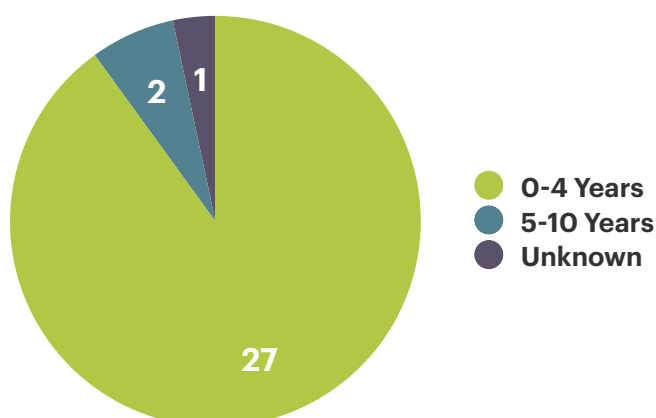
Does the participant experience or has the participant ever experience seizures?

Seizures	Count	Percentage
Yes	30	75%
No	6	15%
Unknown - Not answered	4	10%



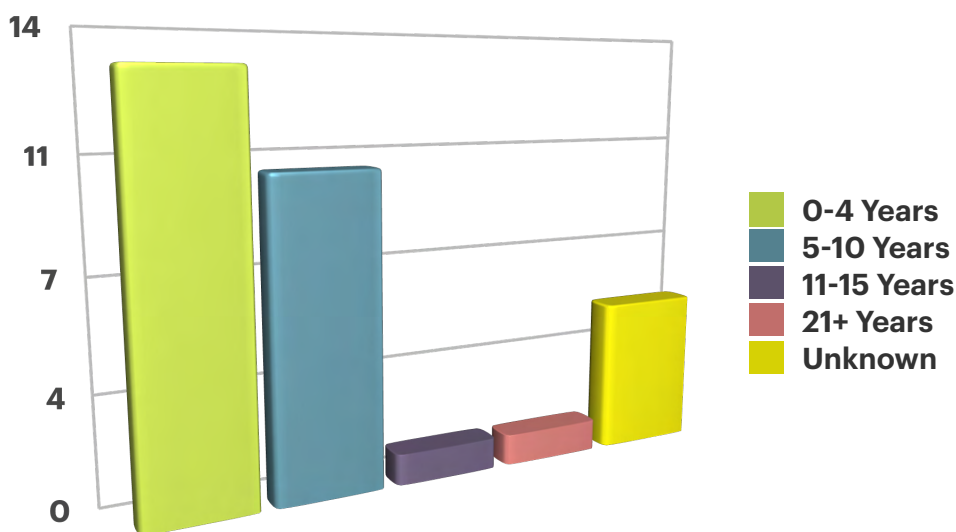
If “yes” at what age did the participant have their first seizure?

Age of first seizure	Count	Percentage
0-4 Years	27	90%
5-10 Years	2	6.67%
Unknown	1	3.33%



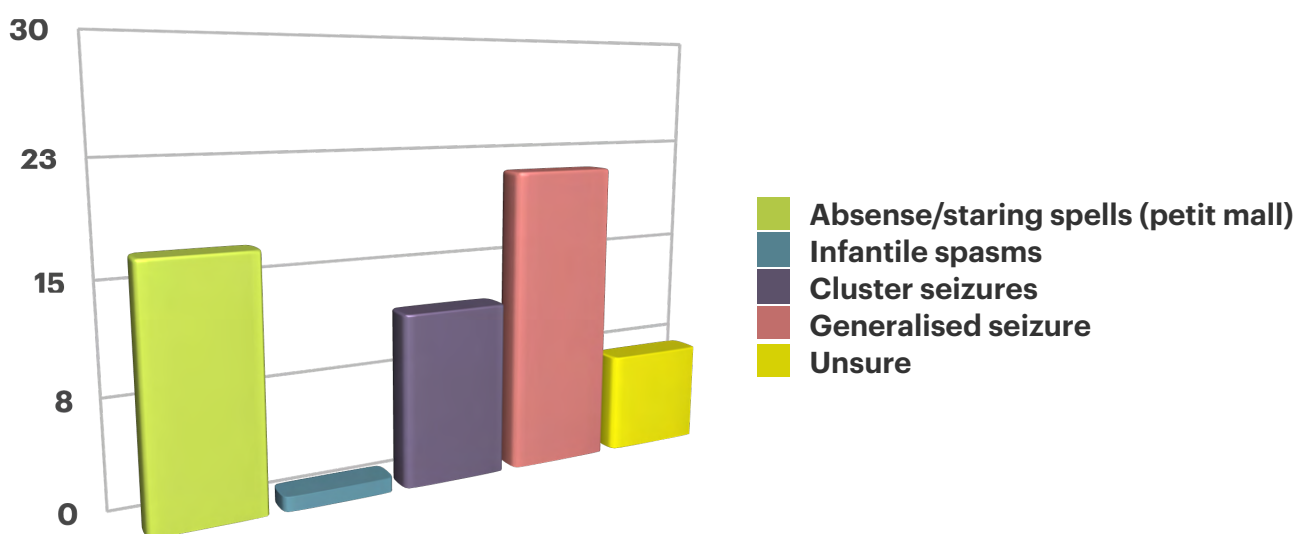
At what age did the participant have their last seizure?

Age of last seizure	Count	Percentage
0-4 Years	13	43.33%
5-10 Years	10	33.33%
11-15 Years	1	3.33%
21+ Years	1	3.33%
Unknown	5	16.68%



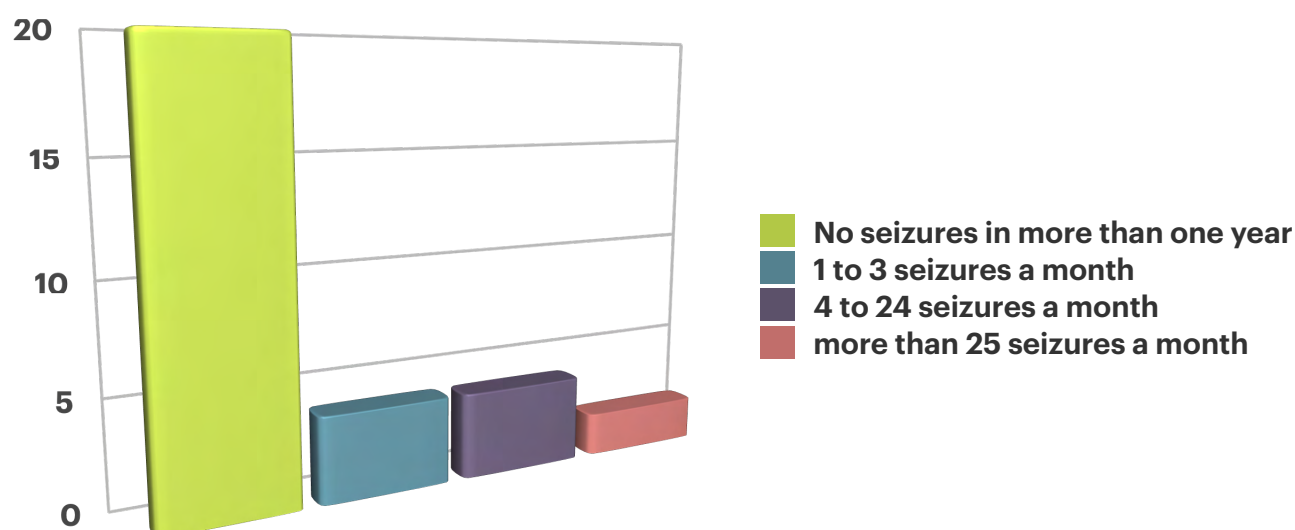
Which of the following best describes the seizures the participant has or experiences?

Type of seizure	Count	Percentage
Absence/staring spells (petit mall)	17	56.66%
Infantile spasms	1	3.33%
Cluster seizures	12	40%
Generalised seizures (tonic clonic, grand mall)	21	70%
Unsure	7	23.33%



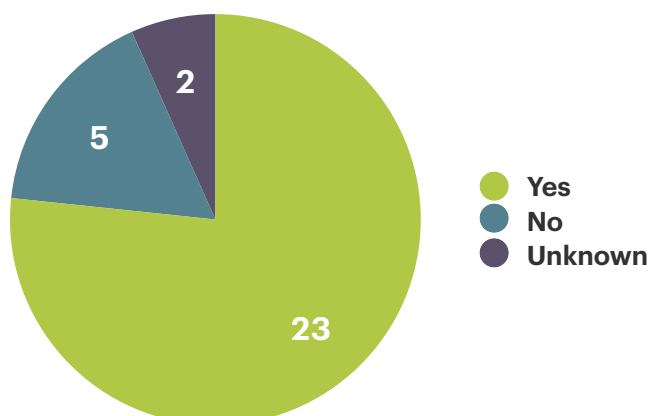
Which of the following best describes how often the participant currently experiences seizures?

Seizure frequency	Count	Percentage
No seizures in more than one year	20	66.66%
1 to 3 seizures a month	4	13.34%
4 to 24 seizures a month	4	13.34%
more than 25 seizure a month	2	6.66%



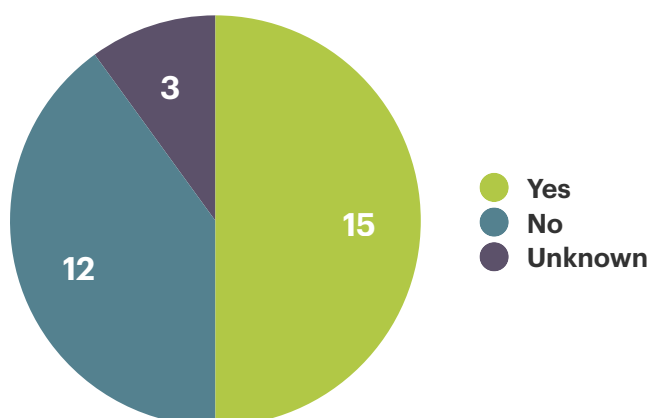
Is the participant currently taking medication to prevent or treat seizures?

Seizure medication	Count	Percentage
Yes	23	76.66%
No	5	16.66%
Unknown	2	6.68%



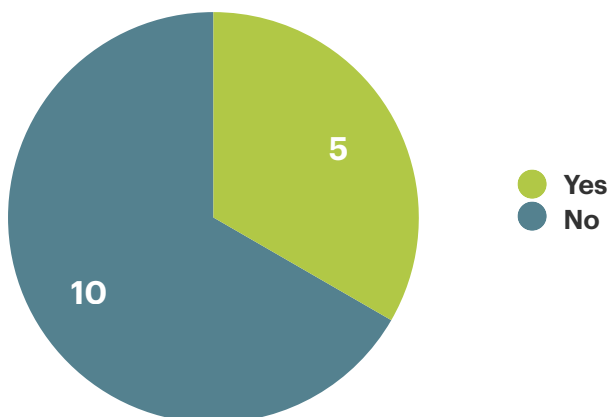
Did the participant ever lose their communication skills after a seizure?

Lose communication	Count	Percentage
Yes	15	50%
No	12	40%
Unknown	3	10%



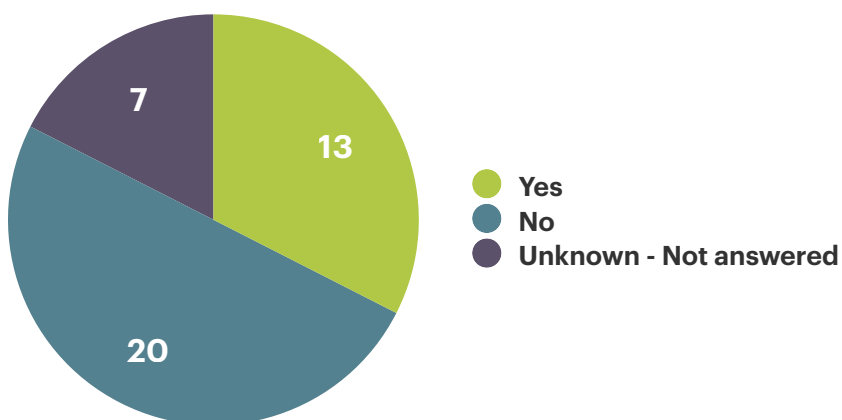
If "yes" did the participant regain their communication skills?

Regained communication	Count	Percentage
Yes	5	33.33%
No	10	66.67%



Does the participant have scoliosis (abnormal curvature of the spine)?

Scoliosis diagnosis	Count	Percentage
Yes	13	32.5%
No	20	50%
Unknown - Not answered	7	17.5%



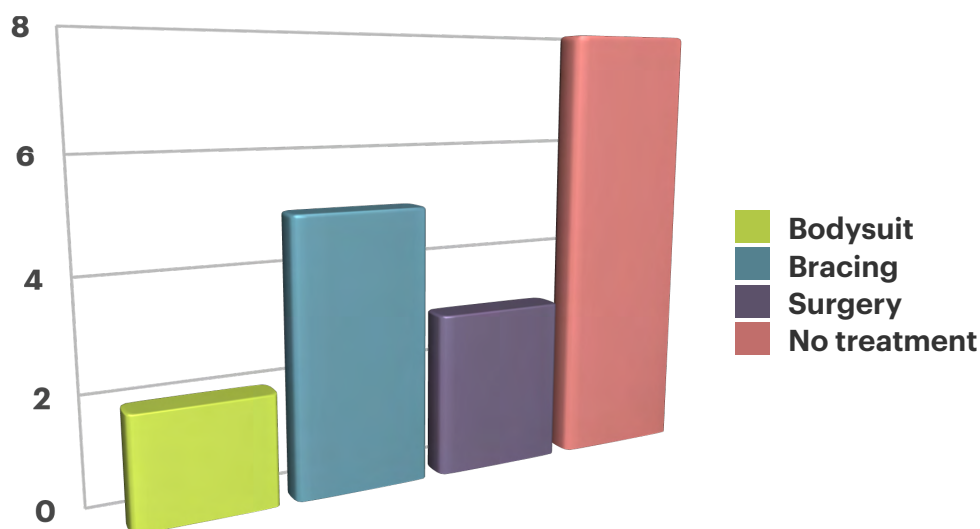
If "yes" how old was the participant when they developed scoliosis?

Age scoliosis diagnosis	Count	Percentage
0-4 Years	4	30.76%
5-10 Years	5	38.46%
11-15 Years	2	15.39%
16-20 Years	2	15.39%



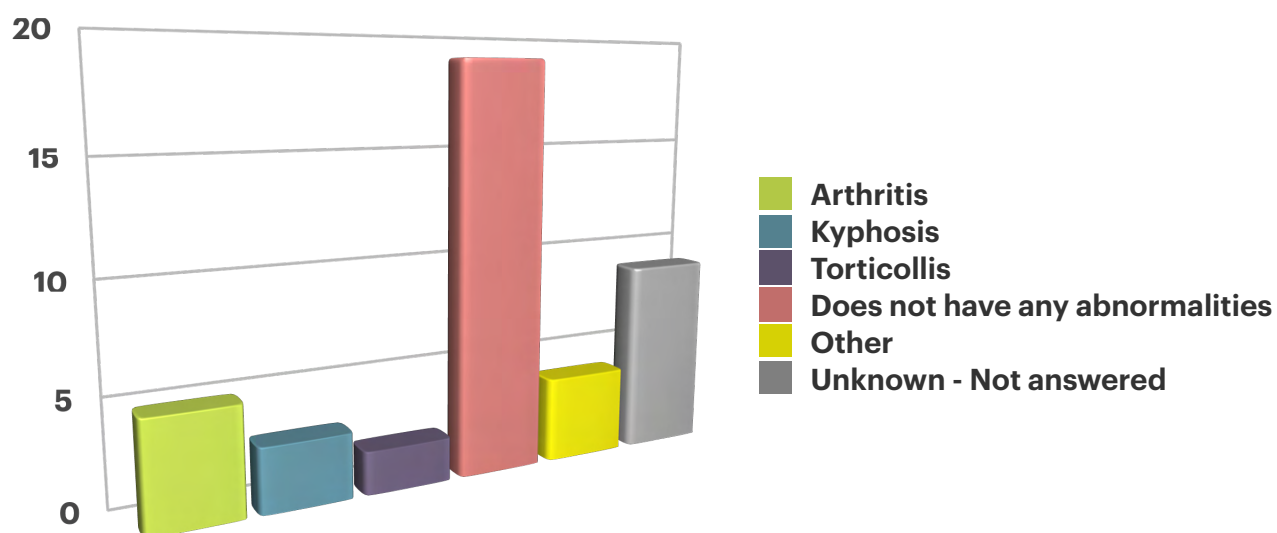
How has the scoliosis been treated?

Scoliosis treatment	Count	Percentage
Bodysuit	2	15.38%
Bracing	5	38.42%
Surgery	3	23.07%
No treatment	8	61.58%



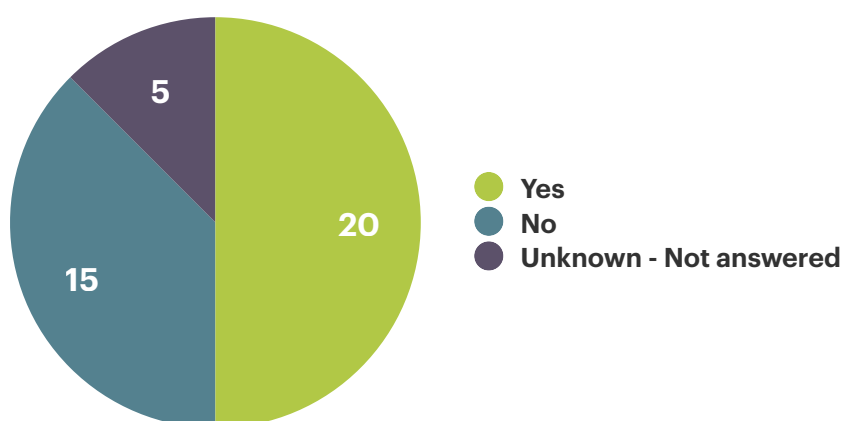
Which of the following best describes the participant’s abnormalities with bone or muscle?

Bone or muscle abnormalities	Count	Percentage
Arthritis	5	12.5%
Kyphosis	3	7.5%
Torticollis	2	5%
Does not have any abnormalities	19	47.5%
Other	4	10%
Unknown - Not answered	9	22.5%



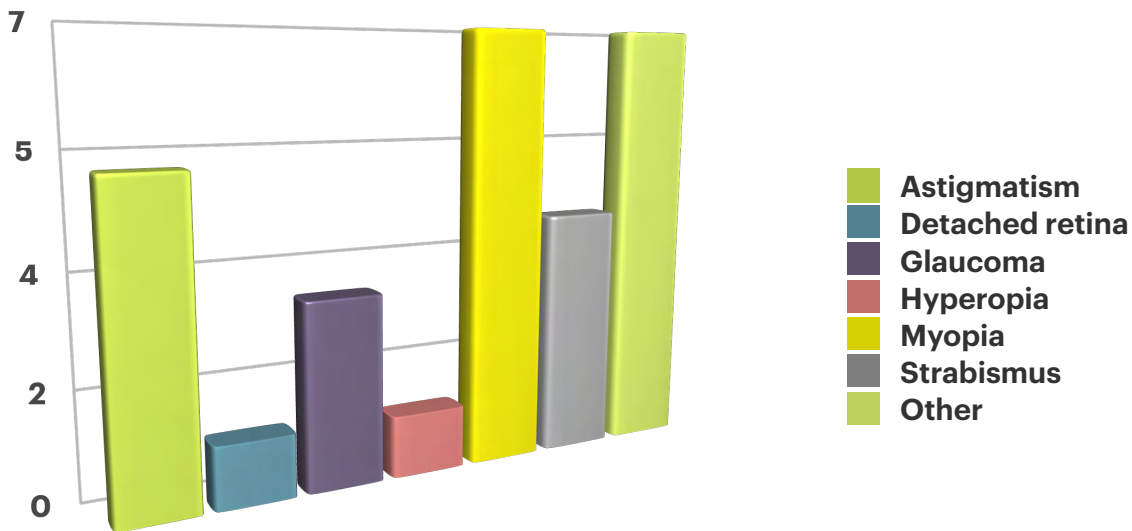
Has the participant ever had any vision or eye problems?

Vision or eye problems	Count	Percentage
Yes	20	50%
No	15	37.5%
Unknown - Not answered	5	12.5%



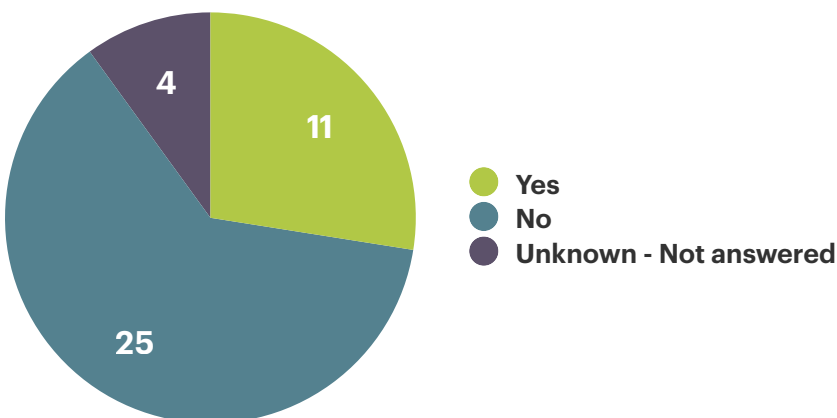
If "yes" which of the following has the participant been diagnosed with?

Vision or eye problem diagnosed	Count	Percentage
Astigmatism	5	25%
Detached retina	1	5%
Glaucoma	3	15%
Hyperopia (far sightedness)	1	5%
Myopia (near sightedness)	7	35%
Strabismus	4	20%
Other	7	35%



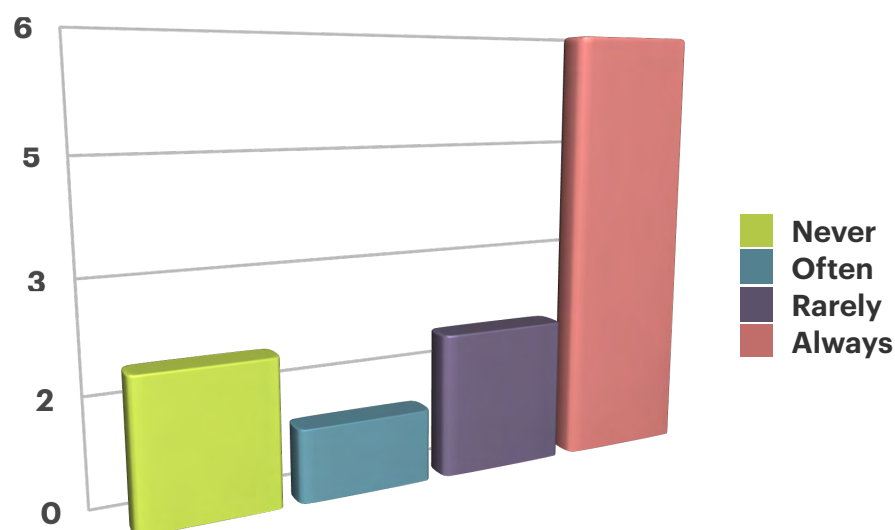
Has the participant been prescribed corrective lenses?

Corrective lenses	Count	Percentage
Yes	11	27.5%
No	25	62.5%
Unknown - Not answered	4	10%



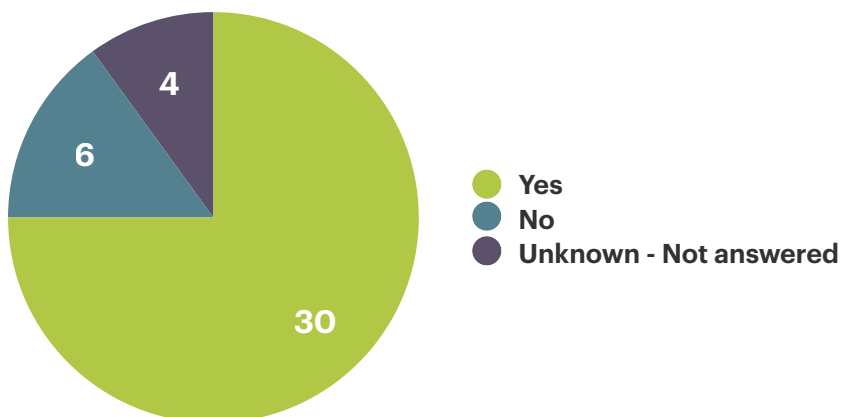
If “yes” how often does the participant wear the corrective lenses?

Length wearing corrective lenses	Count	Percentage
Never	2	18.18%
Often	1	9.09%
Rarely	2	18.18%
Always	6	54.55%



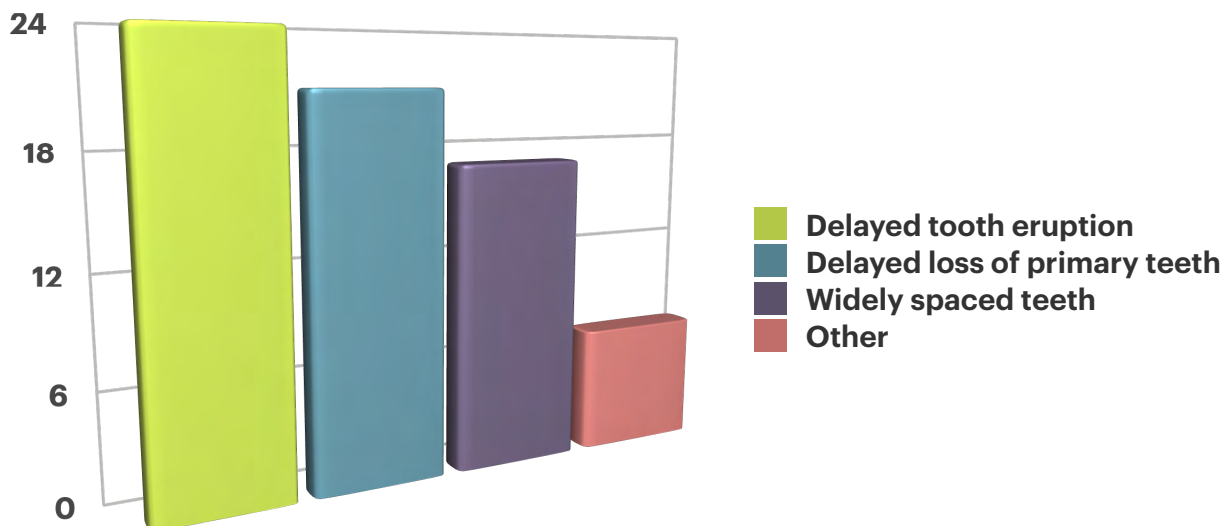
Has the participant ever had any oral/dental issues?

Oral/dental issues	Count	Percentage
Yes	30	75%
No	6	15%
Unknown - Not answered	4	10%



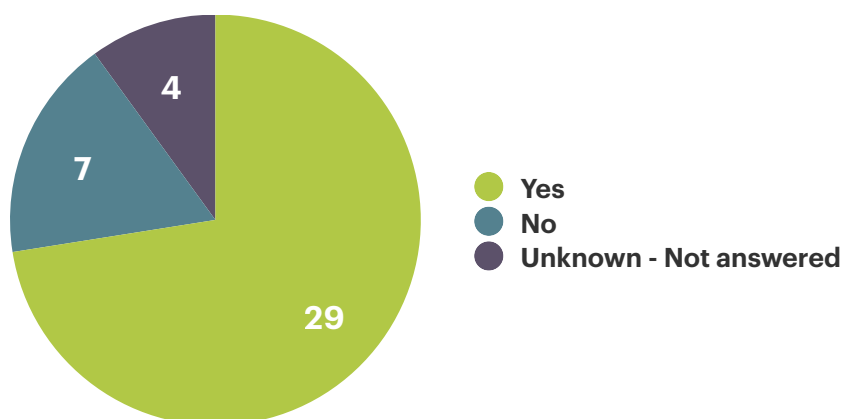
If “yes” which of the following does the participant experience?

Dental issue	Count	Percentage
Delayed tooth eruption	24	80%
Delayed loss of primary teeth	21	70%
Widely spaced teeth	17	56.66%
Other	7	23.33%



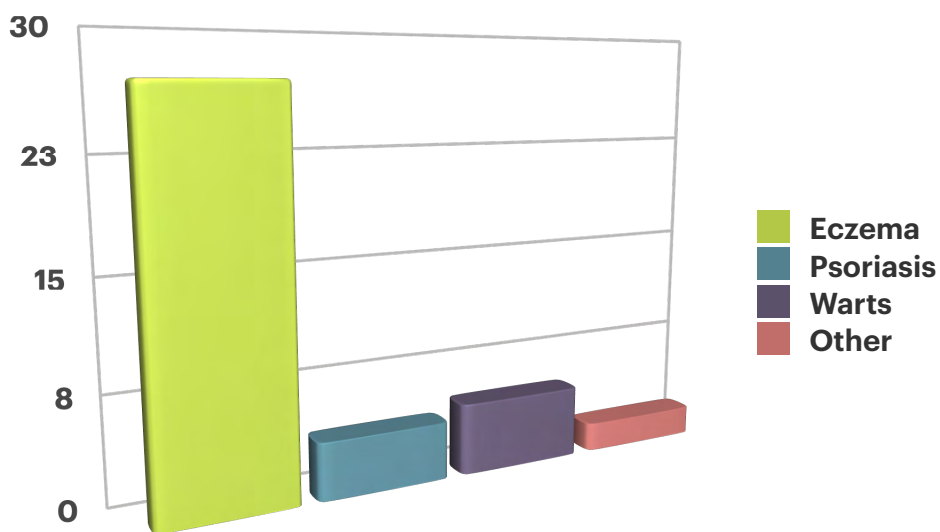
Has the participant ever had any skin issues?

Skin issues	Count	Percentage
Yes	29	72.5%
No	7	17.5%
Unknown - Not answered	4	10%



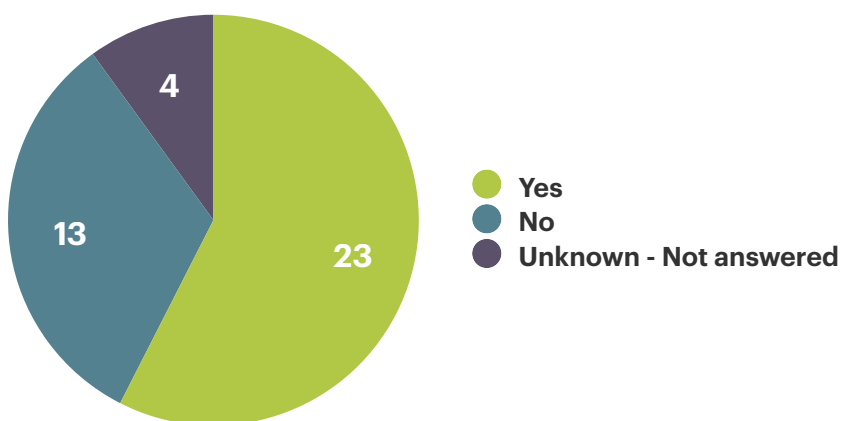
If "yes" which of the following symptoms has the participant experienced?

Skin symptom	Count	Percentage
Eczema	27	93.1%
Psoriasis	4	13.79%
Warts	5	17.24%
Other	2	6.89%



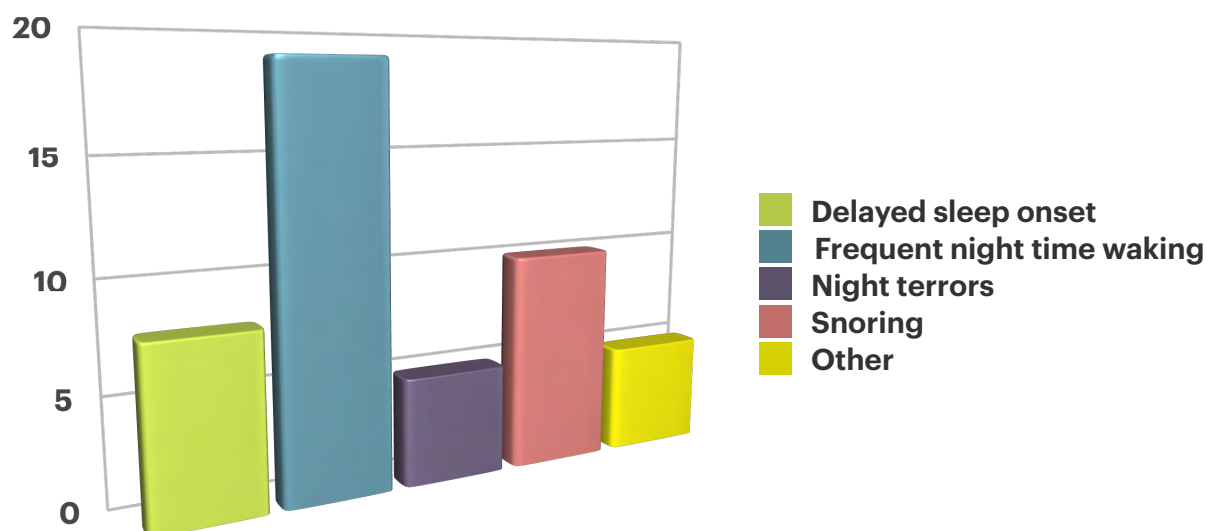
Does the participant have sleep issues?

Sleep issues	Count	Percentage
Yes	23	57.5%
No	13	32.5%
Unknown - Not answered	4	10%



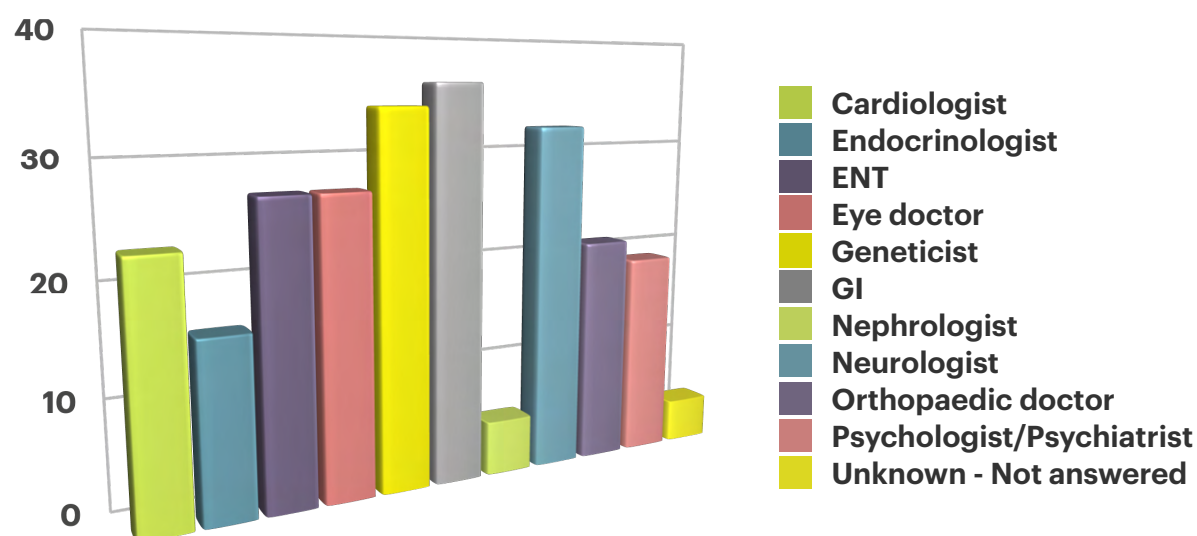
If “yes” which of the following symptoms does the participant experience or has experienced?

Sleep symptom	Count	Percentage
Delayed sleep onset	8	34.78%
Frequent night time waking	19	82.6%
Night terrors	5	21.73%
Snoring	10	43.47%
Other	5	21.73%



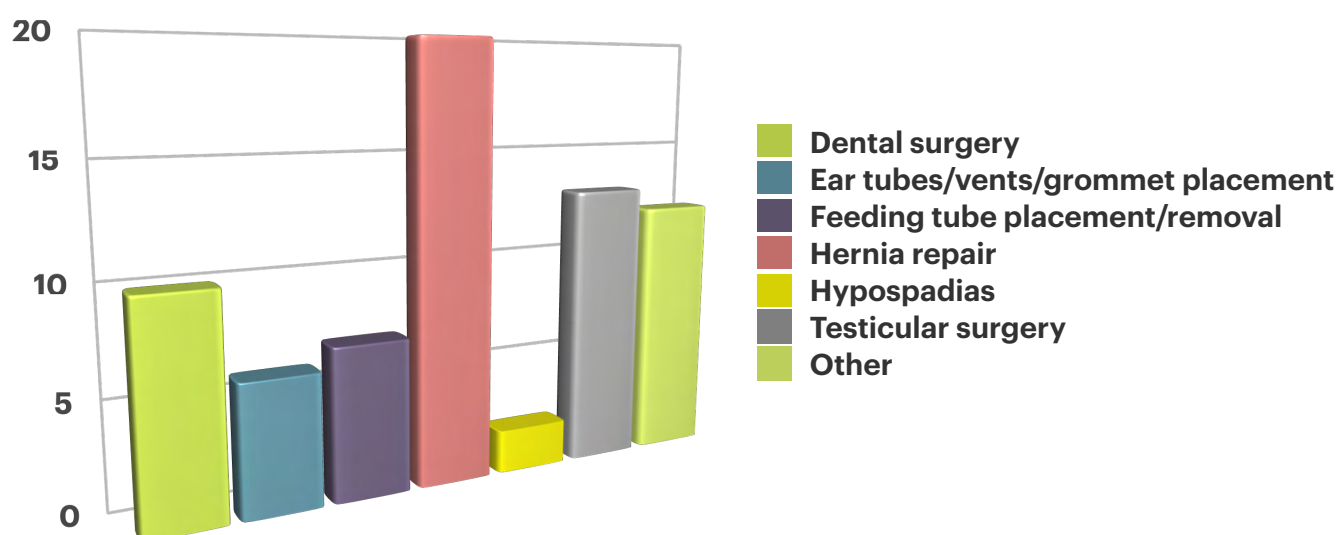
What specialists has the participant seen?

Specialists seen	Count	Percentage
Cardiologist	23	57.5%
Endocrinologist	16	40%
ENT (otolaryngologist)	27	67.5%
Eye doctor (ophthalmologist)	27	67.5%
Geneticist	34	85%
GI (gastroenterologist)	36	90%
Nephrologist	5	12.5%
Neurologist	32	80%
Orthopaedic doctor	21	52.5%
Psychologist/Psychiatrist	19	47.5%
Unknown - Not answered	4	10%



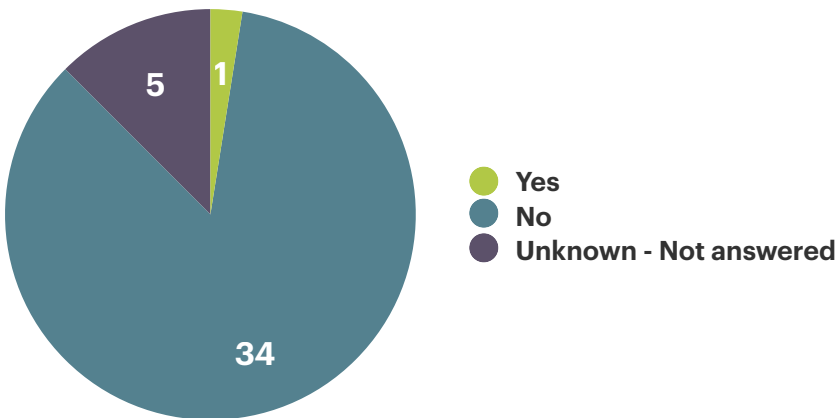
How many surgeries has the participant had?

Surgeries had	Count	Percentage
Dental surgery	10	25%
Ear tubes/vents/grommet placement	6	15%
Feeding tube placement/removal	7	17.5%
Hernia repair	20	50%
Hypospadias	2	5%
Testicular surgery	13	32.5%
Other	12	30%



Has the Participant ever received growth hormone therapy?

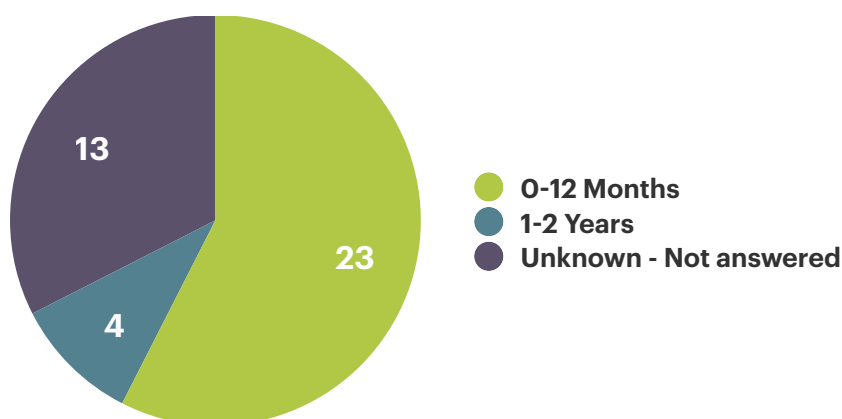
Growth hormone	Count	Percentage
Yes	1	2.5%
No	34	85%
Unknown - Not answered	5	12.5%



Development

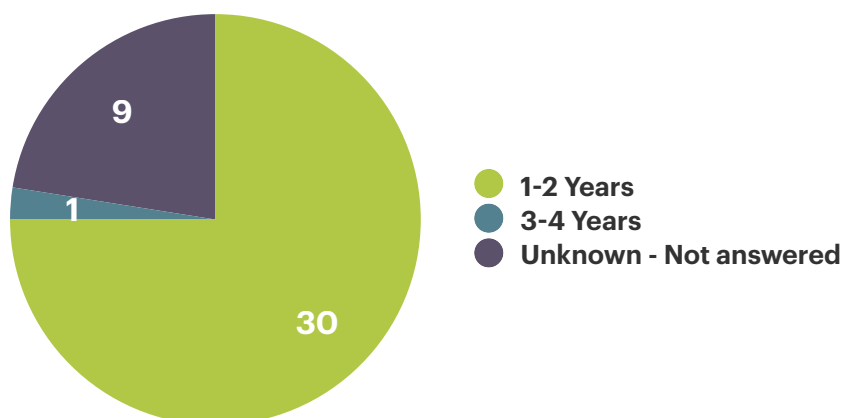
What age was the participant able to sit?

Age able to sit	Count	Percentage
0-12 Months	23	57.5%
1-2 Years	4	10%
Unknown - Not answered	13	32.5%



What age was the participant able to walk?

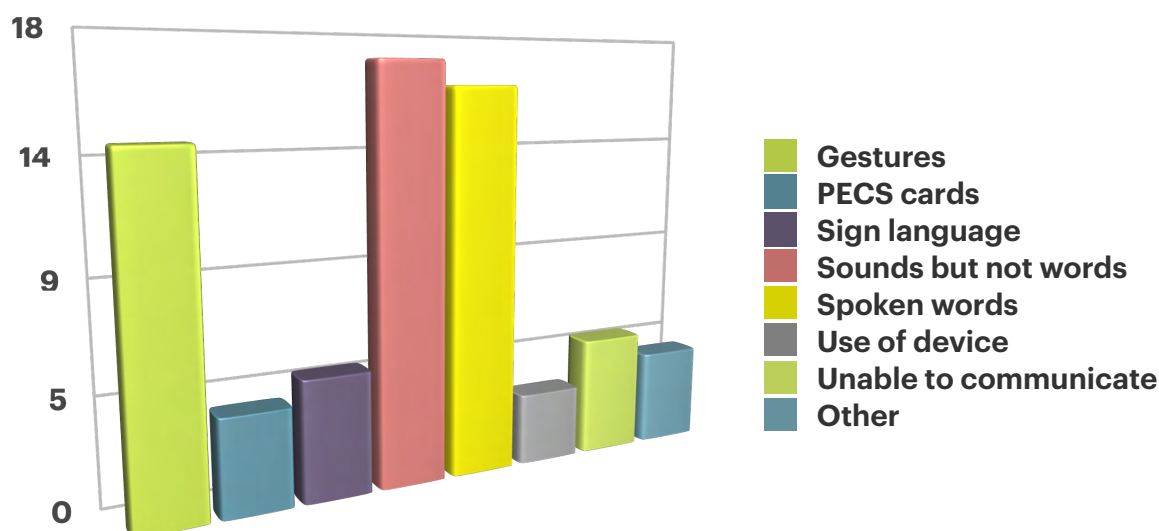
Age able to walk	Count	Percentage
1-2 Years	30	75%
3-4 Years	1	2.5%
Unknown - Not answered	9	22.5%



Communication

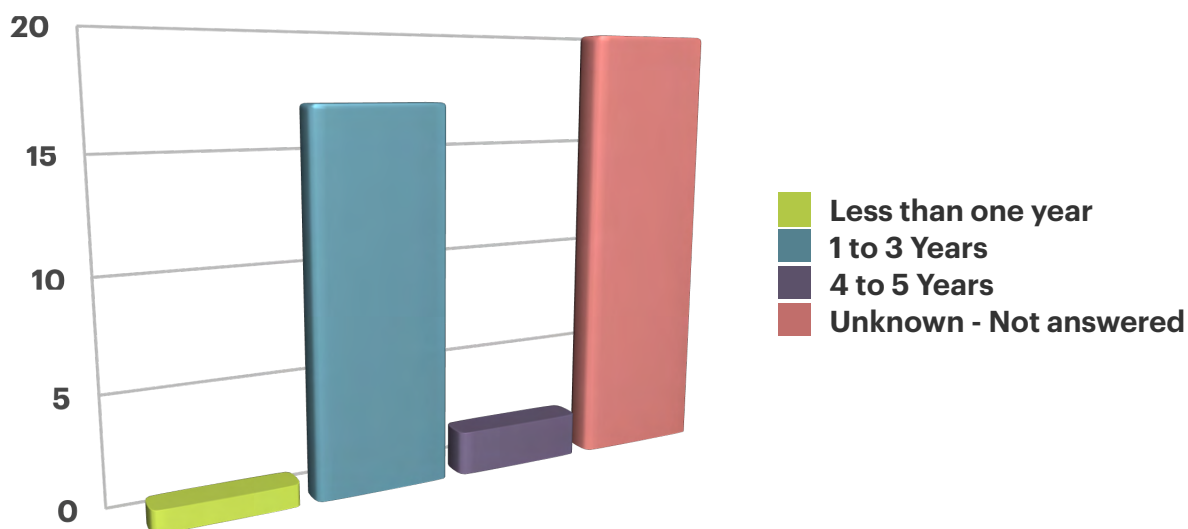
How does the participant communicate?

Communication used	Count	Percentage
Gestures	14	35%
PECS cards (symbol board)	4	10%
Sign language	5	12.5%
Sounds but not words (grunting or laughing)	17	42.5%
Spoken words	16	40%
Use of device (iPad, Proloquo, Dynavox etc.)	3	7.5%
Unable to communicate their wants and needs	5	12.5%
Other	4	10%



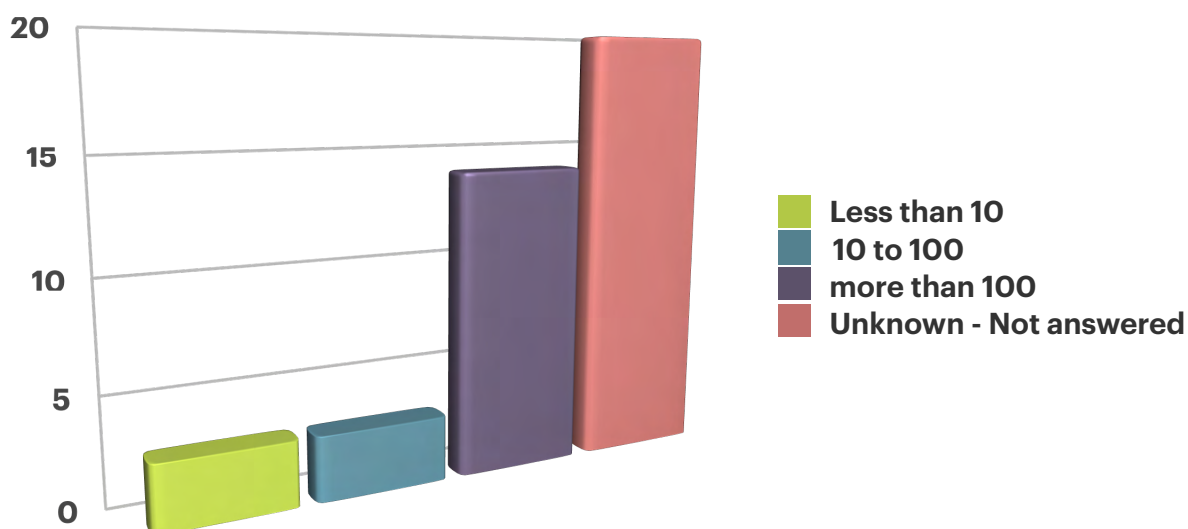
If the participant can speak, how old were they when they spoke their first word?

Words able to speak	Count	Percentage
Less than one year	1	2.5%
1 to 3 Years	17	42.5%
4 to 5 Years	2	5%
Unknown - Not answered	20	50%



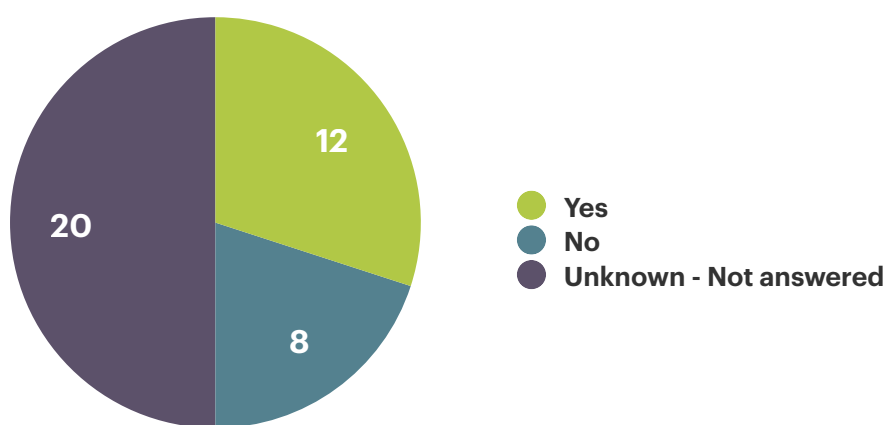
If the participant can speak how many words do they know?

Words able to speak	Count	Percentage
Less than 10	3	7.5%
10 to 100	3	7.5%
more than 100	14	35%
Unknown - Not answered	20	50%



Can the participant speak in full sentences?

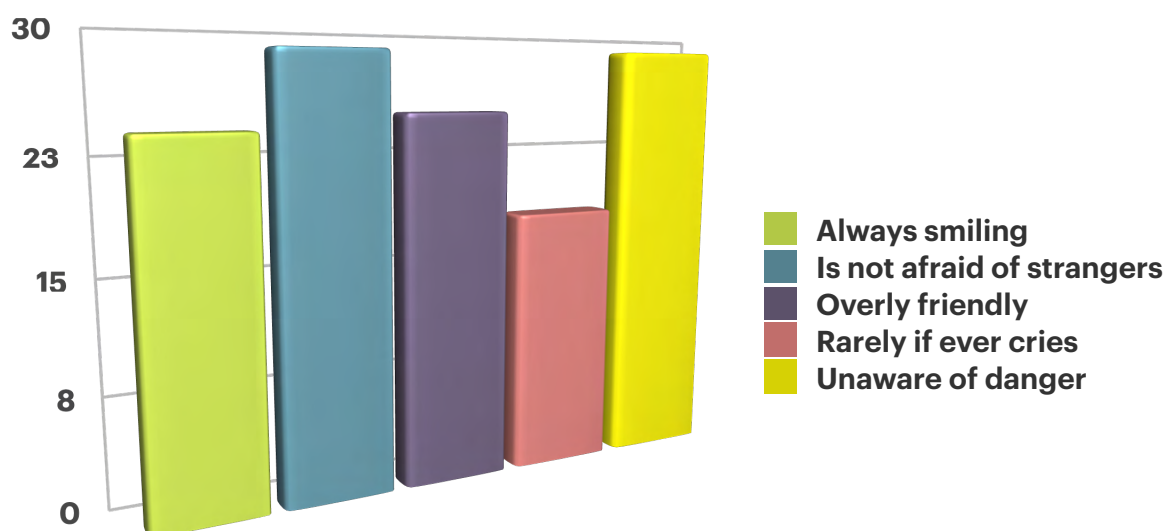
Speak in full sentences	Count	Percentage
Yes	12	30%
No	8	20%
Unknown - Not answered	20	50%



Behaviour

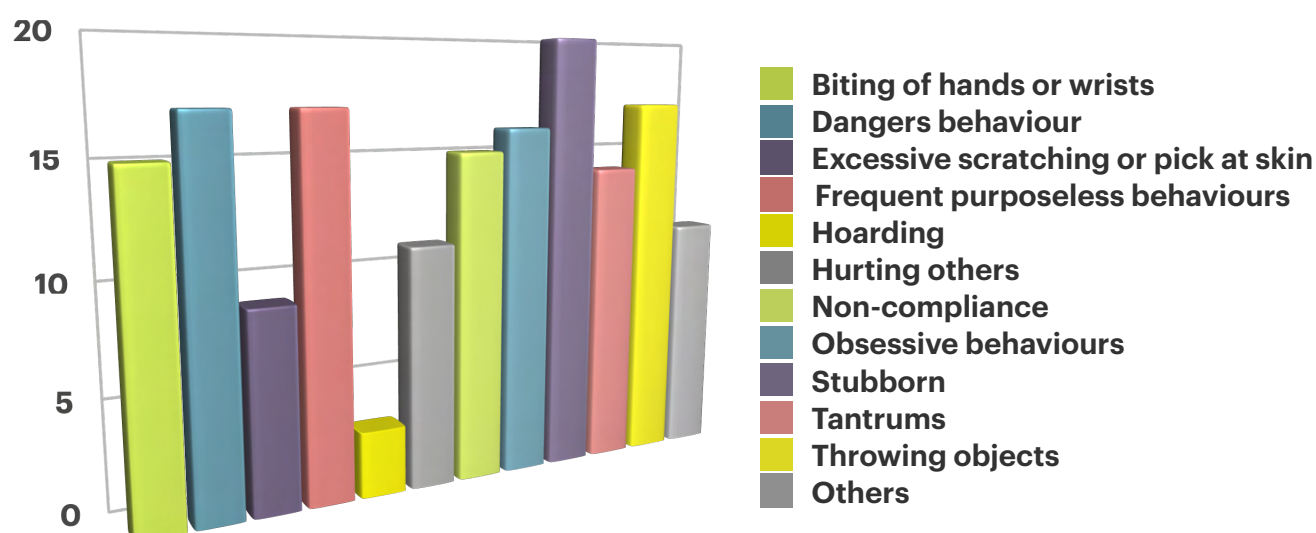
Which of the following traits does the participant currently exhibit?

Traits	Count	Percentage
Always smiling	24	60%
Is not afraid of strangers	29	72.5%
Overly friendly	25	62.5%
Rarely if ever cries	18	45%
Unaware of danger	29	72.5%



Which of the following behavioural issues does the participant currently exhibit?

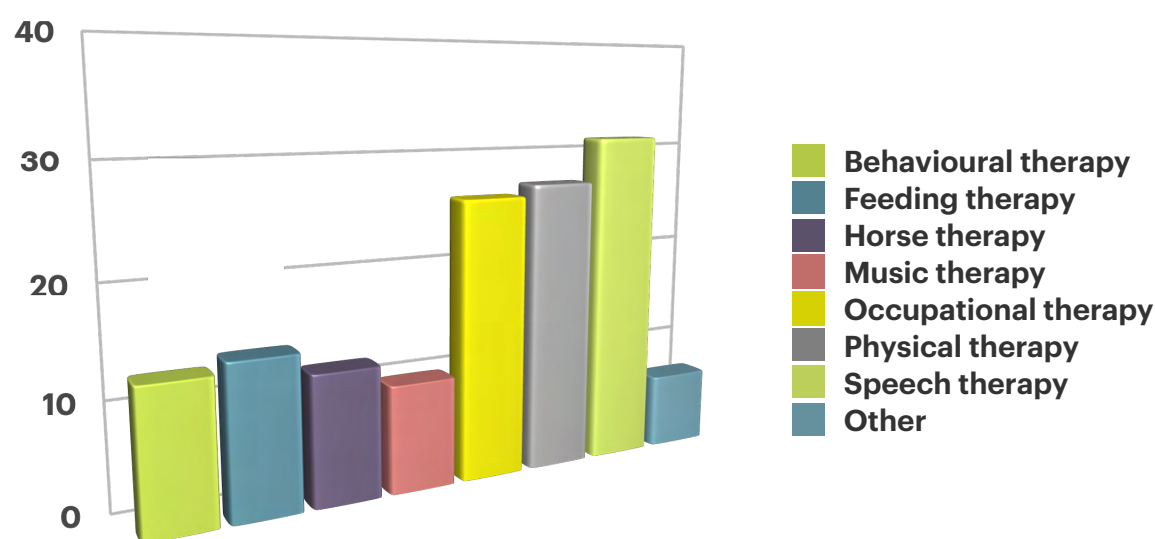
Behavioural issue	Count	Percentage
Biting of hands or wrists	15	37.5%
Dangers behaviour (such as running away or dropping to the ground)	17	42.5%
Excessive scratching or pick at skin	9	22.5%
Frequent purposeless behaviours (hand flapping, tapping objects, spinning objects)	17	42.5%
Hoarding	3	7.5%
Hurting others (Scratching, hitting, pinching, biting)	11	27.5%
Non-compliance (refusing to follow important instructions)	15	37.5%
Obsessive behaviours	16	40%
Stubborn	20	50%
Tantrums	14	35%
Throwing objects	17	42.5%
Others	11	27.5%



Therapy

What type of services/therapy has the participant received?

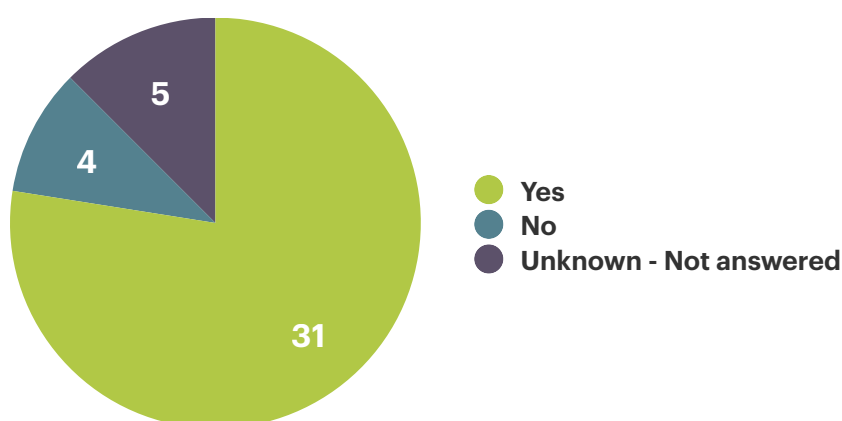
Services/therapy	Count	Percentage
Behavioural therapy	13	32.5%
Feeding therapy	14	35%
Horse therapy	12	30%
Music therapy	10	25%
Occupational therapy	26	65%
Physical therapy	27	67.5%
Speech therapy	31	77.5%
Other	7	17.5%



Education

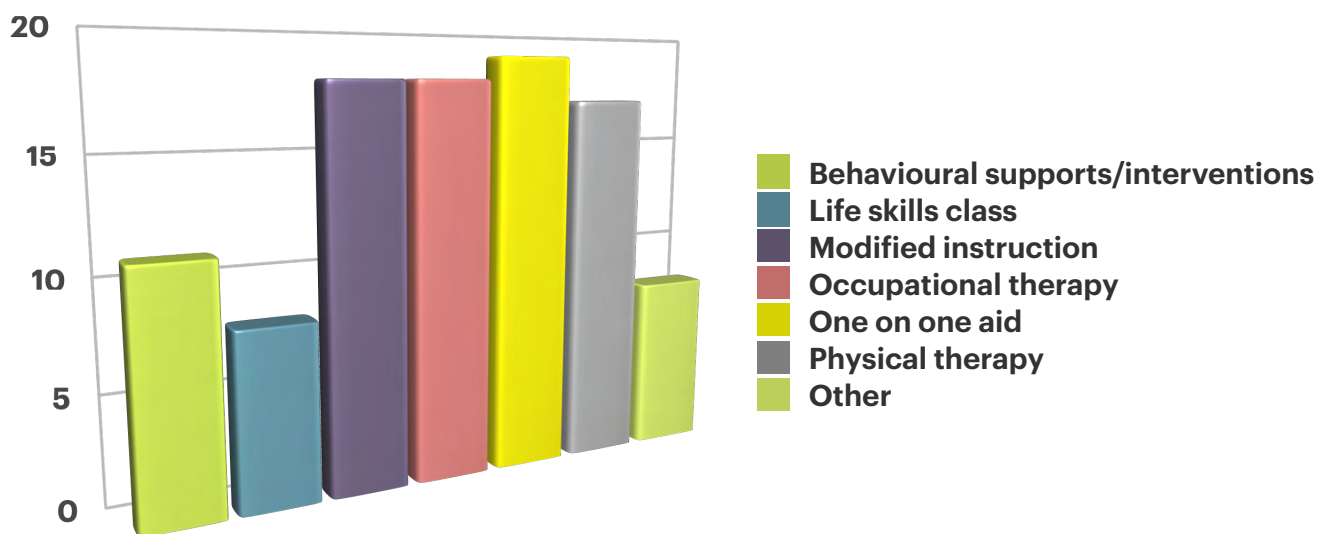
Is the participant currently in school or attended school in the past?

Attend school	Count	Percentage
Yes	31	77.5%
No	4	10%
Unknown - Not answered	5	12.5%



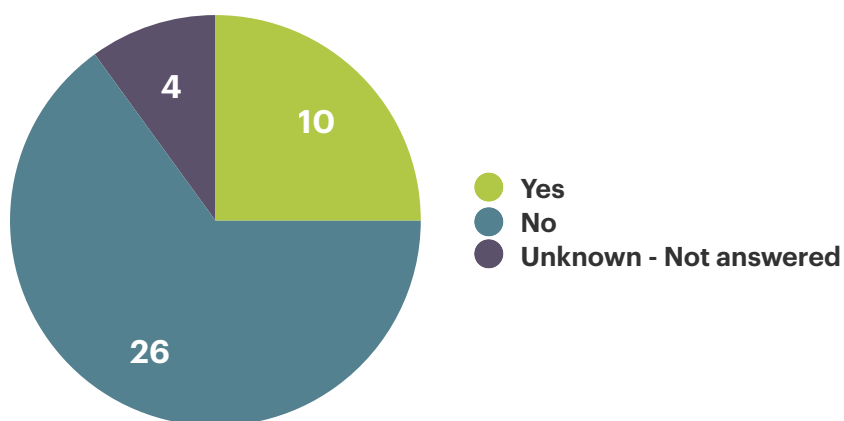
If the participant received services/supports, what type of school services/supports has the participant received upon entering the school system?

Services/supports	Count	Percentage
Behavioural supports/interventions	11	27.5%
Life skills class	8	20%
Modified instruction	18	45%
Occupational therapy	18	45%
One on one aid	19	47.5%
Physical therapy	17	42.5%
Other	8	20%



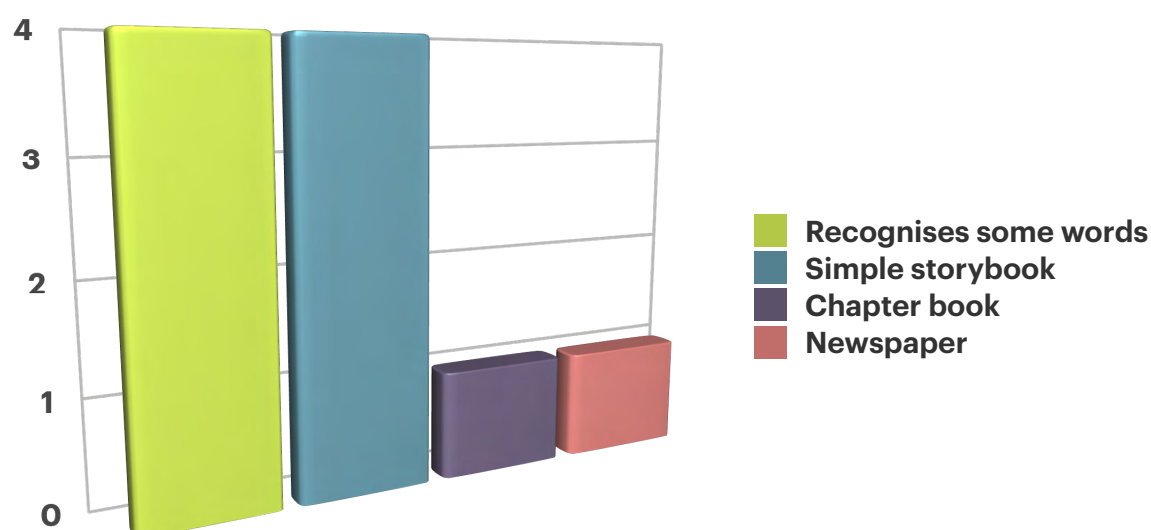
Is the participant able to read?

Can participant read	Count	Percentage
Yes	10	25%
No	26	65%
Unknown - Not answered	4	10%



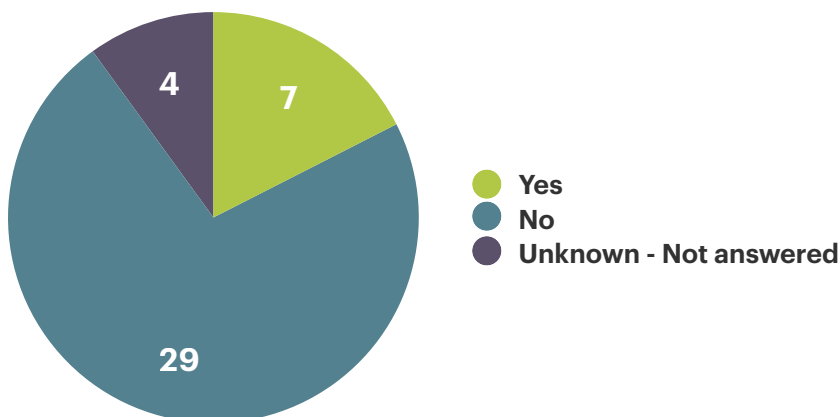
If “yes” which of the following best describes the participants reading ability?

Reading ability	Count	Percentage
Recognises some words (such as their name)	4	40%
Simple storybook	4	40%
Chapter book	1	10%
Newspaper	1	10%



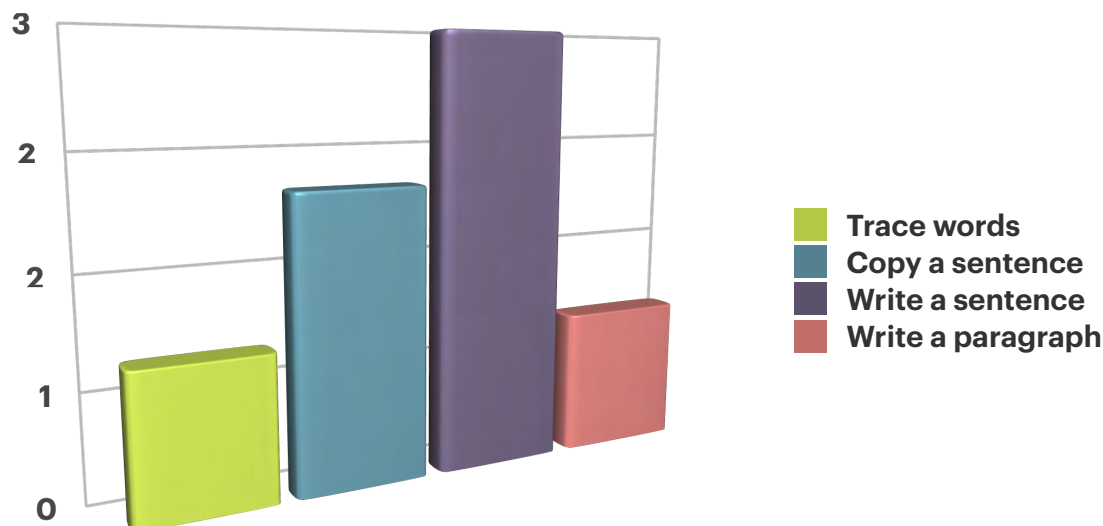
Is the participant able to write?

Can participant read	Count	Percentage
Yes	7	17.5%
No	29	72.5%
Unknown - Not answered	4	10%



If “yes” which of the following best describes the participants writing ability?

Reading ability	Count	Percentage
Trace words	1	14.28%
Copy a sentence	2	28.56%
Write a sentence independently	3	42.88%
Write a paragraph	1	14.28%



This concludes the report. We thank you for taking the time to look through, we hope this will help you to better understand NCBRS that bit better.



Acknowledgments

We would like to thank all the NCBRS patients and their parent/guardian's for taking the time to enrol and fill in the NCBRS Patient Registry questionnaire. We encourage more families to enrol to help us all better understand NCBRS.

We would also like to thank the whole CoRDS team for giving us the opportunity to establish and create a global NCBRS Patient Registry. We are already able to gather the much needed data to enable our patients, their families and potential researchers to better understand this rare condition.

Thank You

Yours sincerely

Lee Reavey

Helen Robinson

Co-Founder/CEO

Co-Founder/Chair