



# Kleefstra Syndrome Survey Results 2018

Summer 2018

# INTRODUCTION

The Kleefstra syndrome parent survey was carried out in June 2018 and was distributed over social media for parents or primary carers of people diagnosed with Kleefstra syndrome to complete. There were 179 respondents from 22 different countries, regarding people with Kleefstra syndrome ranging from under 12 months up to 38 years old.

Kleefstra syndrome is a rare genetic condition caused by a deletion or mutation to the EHMT1 gene towards the end of the 9th chromosome. This gene is responsible for the activation of other genes, so a malfunctioning EHMT1 gene can affect multiple organs and/or functions.

The aim of this short survey was to try to provide some answers to those common questions that pop up fairly frequently especially for those with a new diagnosis, such as how old was your child when they started to walk, how many children also have an autism spectrum diagnosis, and so on.

It is important to remember that there is a large spectrum of symptoms in Kleefstra syndrome, and no two children are the same. Everyone develops at their own rate. Consult with professionals for specific medical or therapeutic advice as required. And do not give up!

We should also note that due to the rare nature of the syndrome, the sample size completing the survey is small and so results may not be generalisable. Some questions are reliant on people's memories which may also affect reliability.

We are grateful to all the families across our community that took the time to complete this survey – thank you.

## KEY FACTS

### **Diagnosis**

- Nearly 50% were diagnosed with Kleefstra syndrome before they were 3.
- 7% got their diagnosis over the age of 18.

### **Key symptoms**

There was a range of key symptoms or additional diagnoses, the five listed below were most prevalent (followed by the percentage of respondents who said their child experienced or was diagnosed with it):

- Constipation (60%)
- Autism Spectrum Condition (40%)
- Aspiration Reflux/GERD (38%)
- Hearing impairment (38%)
- Heart condition (36%)

7% of respondents said their child was not affected by any of the listed symptoms.

### **School**

- The most popular educational setting was a special school; over a third of children attend (or attended) a special school.

## **Getting around**

- 57% of people walk without additional equipment.
- 16% are wheelchair or buggy dependent.
- Of those that took the survey, 85% were deemed able to walk by the respondent, all by the age of 14. Over half of the children that are able to walk were walking by the age of 2 (55%).

## **Toileting**

- 57% of those surveyed are not toilet trained.
- 43% are toilet trained to some level, and just 18% are dry both day and night.
- Of those that are toilet trained, 49% were toilet trained by the age of 4, and all were toilet trained by the age of 13.

## **Communication**

- 29% are classed as non-verbal without using additional communication methods, and on the flip side, 16% talk in full sentences. The rest use various methods of communication with differing success, the most popular being sign language.
- 44% deemed their child able to talk coherently/using sentences, and all of those were talking before 11. Just over a third of those that can talk were talking before they were 5.

## **Regression**

- 21% of those completing the survey had experienced regression or were experiencing it at the moment; 79% have not experienced regression.
- We didn't ask the age that the previous regression had occurred but looking at current ages they tend to be in the teen/adolescence age or older. Interestingly, there are some people with Kleefstra syndrome in their 20s and 30s who have not experienced regression, but sample sizes are very small to be able to gauge the significance of this.

## **Other**

- 42% of people with Kleefstra syndrome wake frequently during the night.
- 41% display anxiety to an excessive degree.
- 37% are very affectionate and 27% laugh a lot!
- 17% display self harming behaviour.

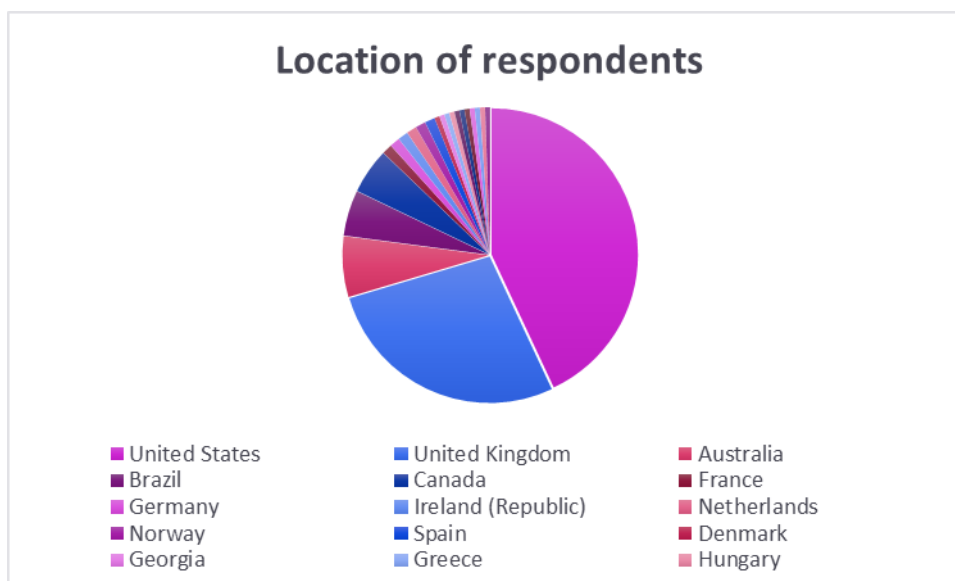
# FULL RESULTS

## LOCATION

In total, 179 families completed this survey, from 22 different countries. The table and chart below shows the countries from which the completions were from.

Question: Where in the world are you?

	Number	Percentage
United States	77	43%
United Kingdom	49	27%
Australia	12	7%
Brazil	9	5%
Canada	9	5%
France	2	1%
Germany	2	1%
Ireland (Republic)	2	1%
Netherlands	2	1%
Norway	2	1%
Spain	2	1%
Denmark	1	1%
Georgia	1	1%
Greece	1	1%
Hungary	1	1%
Iceland	1	1%
Italy	1	1%
New Zealand	1	1%
Russian Federation	1	1%
South Africa	1	1%
Sweden	1	1%
Turkey	1	1%
<i>Total</i>	<i>179</i>	

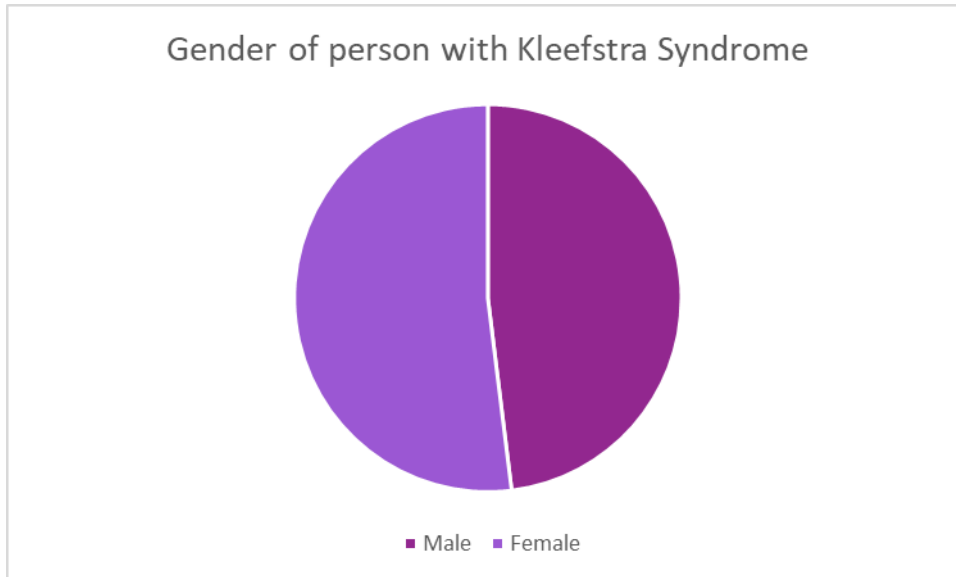


## GENDER

The gender split is 48% male and 52% female.

The gender split for UK based families was 63% male and 37% female – in the US, the split was the other way; 42% male and 58% female.

Question: What sex is your child?

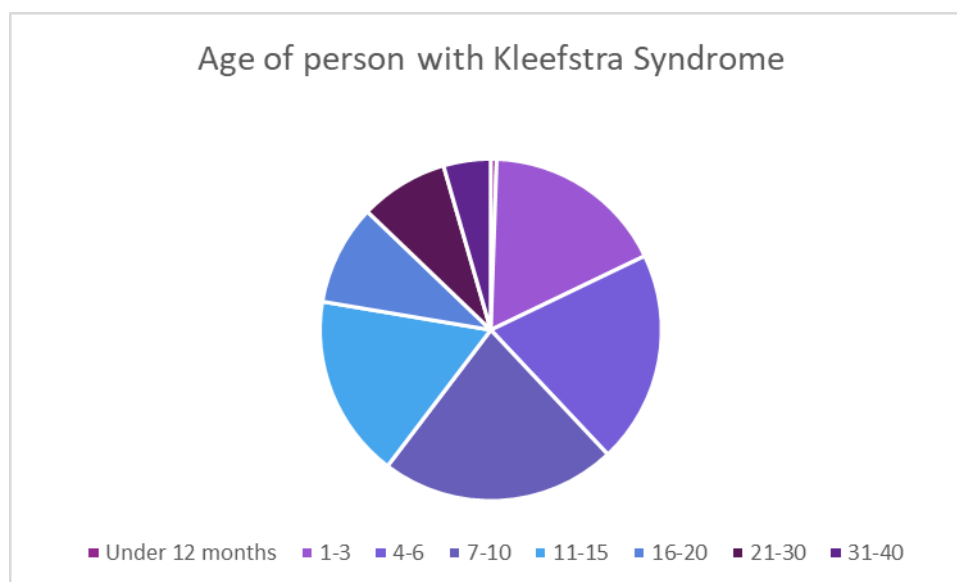


## AGE

The ages ranged from under 12 months up to 38 years old, with 82% of completions from families with children under the age of 18 (as of 1<sup>st</sup> August 2018). The eldest in the survey were two people aged 38, from Canada and Australia.

Question: How old will your child be on 1<sup>st</sup> August 2018?

	<i>Number</i>	<i>Percentage</i>
Under 12 months	1	1%
1-3	31	17%
4-6	36	20%
7-10	40	22%
11-15	31	17%
16-20	17	9%
21-30	15	8%
31-40	8	4%
<i>Total</i>	<i>179</i>	

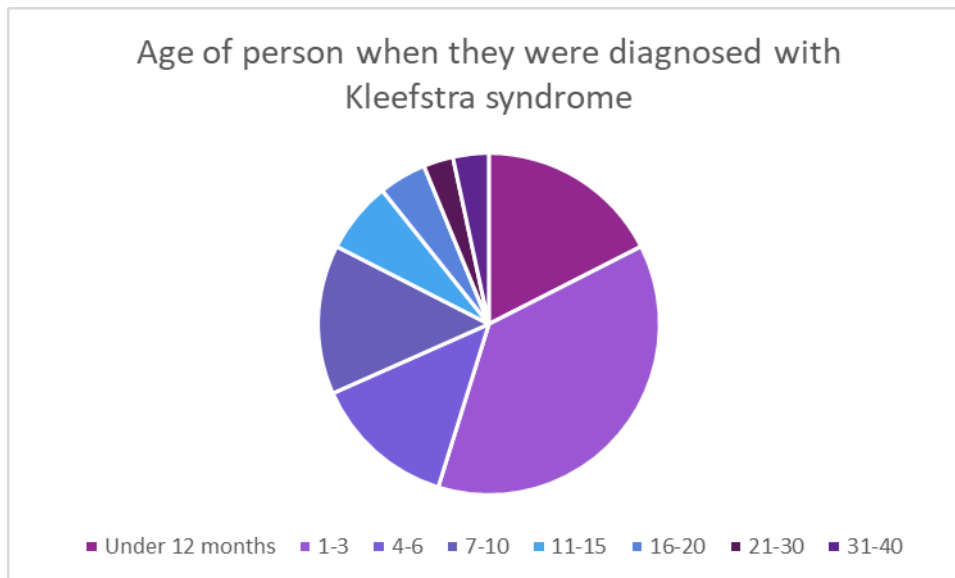


## AGE OF DIAGNOSIS

There is a fairly broad spread of ages for diagnosis, going right up into the thirties. Of those who could remember the age of their child when they were diagnosed (2 couldn't), 49% were diagnosed before they were 3. 7% were diagnosed over the age of 18.

Question: At what age was your child diagnosed?

	Number	Percentage
Not sure	2	-
Under 12 months	31	18%
1-3	66	37%
4-6	24	14%
7-10	25	14%
11-15	12	7%
16-20	8	5%
21-30	5	3%
31-40	6	3%
Total	179	

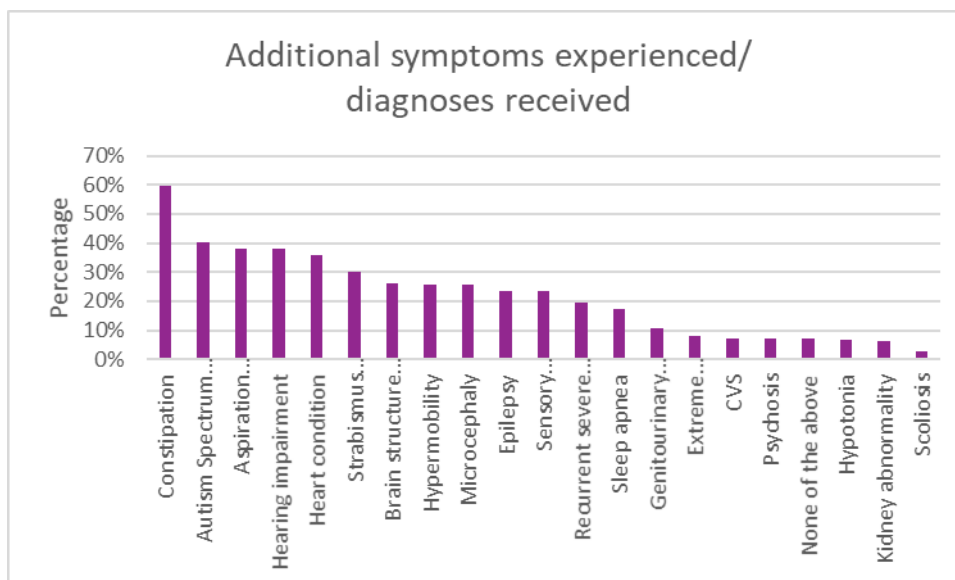


## KEY SYMPTOMS/ADDITIONAL DIAGNOSES

The full list of things people and have been diagnosed with or experienced is below, along with prevalence:

Question: Have they also been diagnosed with or experienced any of the following?

	<i>Number affected</i>	<i>Percentage</i>
Constipation	107	60%
Autism Spectrum Condition	72	40%
Aspiration Reflux/GERD	68	38%
Hearing impairment	68	38%
Heart condition	64	36%
Strabismus (squint) or other unusual eye features	54	30%
Brain structure abnormalities	47	26%
Hypermobility	46	26%
Microcephaly	46	26%
Epilepsy	42	23%
Sensory Integration Disorder	42	23%
Recurrent severe respiratory/pulmonary infections	35	20%
Sleep apnea	31	17%
Genitourinary Abnormality	19	11%
Extreme apathy/catatonia	14	8%
Cyclic Vomiting Syndrome (CVS)	13	7%
Psychosis	13	7%
None of these options	13	7%
Hypotonia	12	7%
Kidney abnormality	11	6%
Scoliosis	5	3%





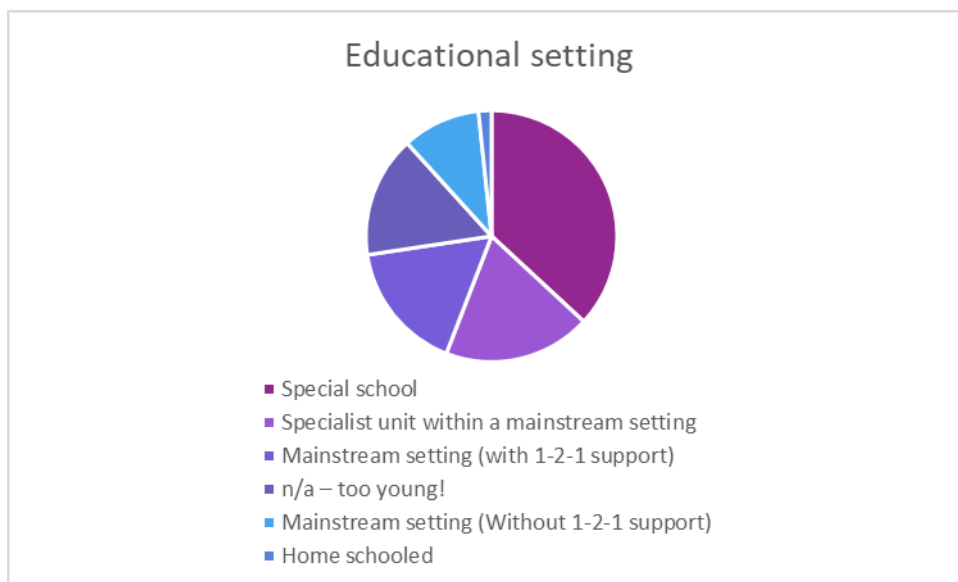
## EDUCATIONAL SETTING

Over a third of children attend or attended for the majority of their schooling a special school. The next most popular was a specialist unit within a mainstream setting, followed by mainstream setting with 1-2-1 support, then

All of those home schooled (3) were based in the US.

Question: What kind of educational setting does your child currently attend/did your child attend for the majority of their schooling?

	<i>Number</i>	<i>Percentage</i>
Special school	66	37%
Specialist unit within a mainstream setting	34	19%
Mainstream setting (with 1-2-1 support)	30	17%
n/a – too young!	28	16%
Mainstream setting (Without 1-2-1 support)	18	10%
Home schooled	3	2%
<i>Total</i>	<i>179</i>	

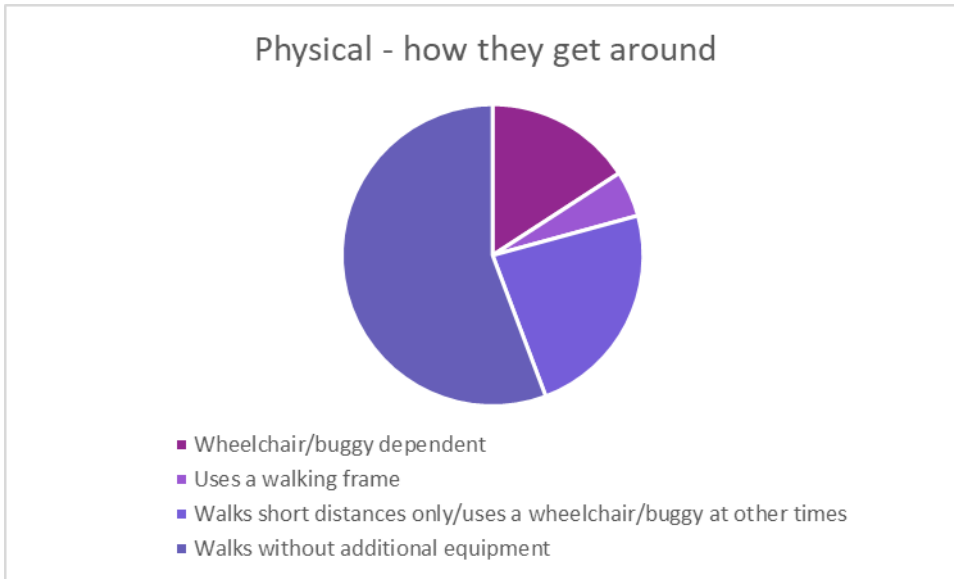


## GETTING AROUND

Over half walk without additional equipment and, at the other end, 16% are wheelchair or buggy dependent. (People were able to select more than one option here.)

Question: How does your child get around?

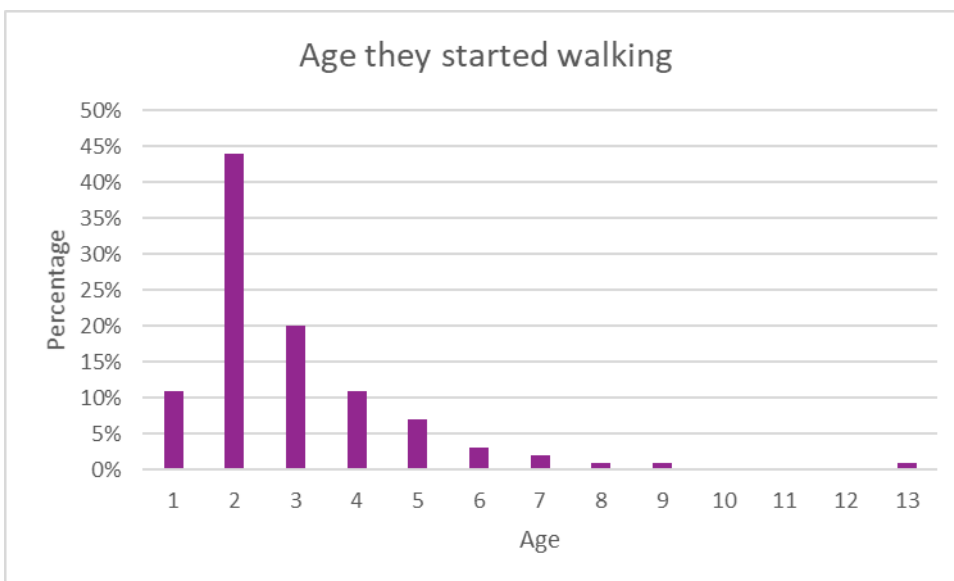
	Number	Percentage
Wheelchair/buggy dependent	29	16%
Uses a walking frame	9	5%
Walks short distances only/uses a wheelchair/buggy at other times	43	24%
Walks without additional equipment	102	57%



## Walking – Age

Of all those that took the survey, 85% are deemed able to walk, all by the age of 14. Over half of the children that are able to walk were walking by the age of 2 (55%).

Question: If your child can walk, at what age would you say they started to walk?



## TOILET TRAINING

43% are toilet trained to some level, and just 18% of those are dry during the day and the night. 57% are not toilet trained.

Question: Is your child toilet trained?

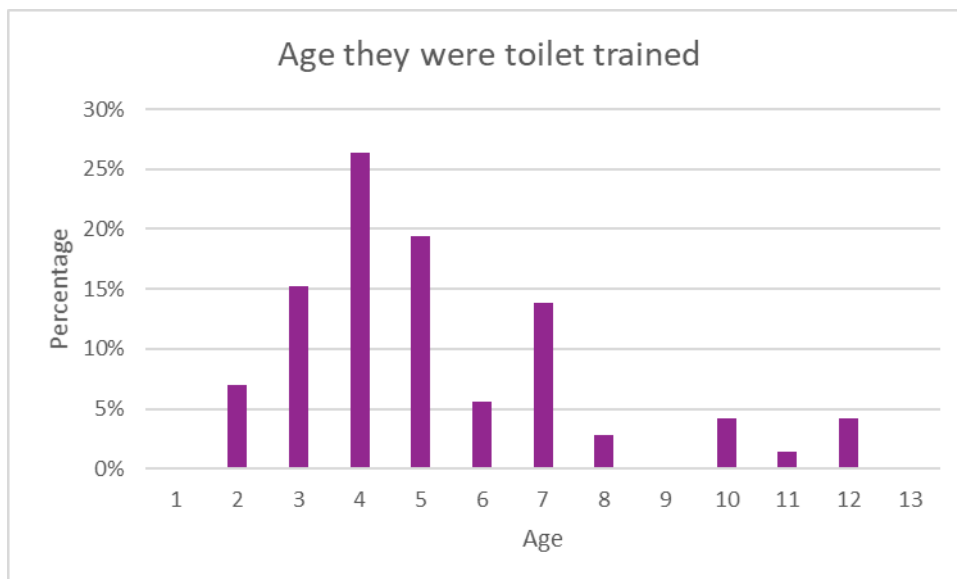
	Number	Percentage
No – uses nappies, pads or continence products	102	57%
Yes – dry during the day, uses nappies, pads or continence products at night	44	25%
Yes – dry during both day and night	33	18%
Total	179	



### Toileting – Age

Of those that are toilet trained, 49% were toilet trained by the age of 4, and all were toilet trained by the age of 13.

Question: If your child is potty trained, at what age would you say this occurred?

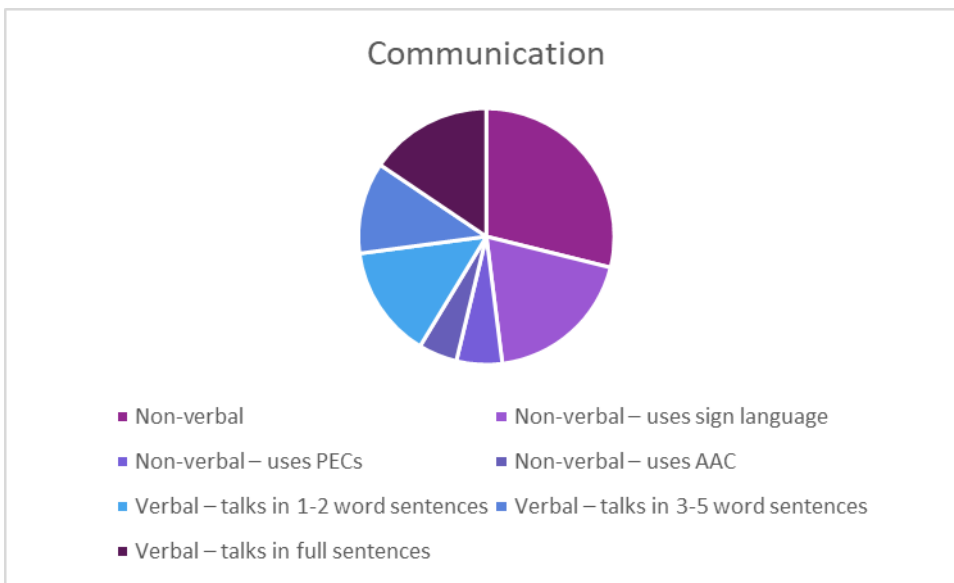


## COMMUNICATION

29% are classed as non-verbal without using additional communication methods, and on the flip side, 16% talk in full sentences. The rest use various methods of communication with different success, the most popular being sign language. (People were able to select more than one option here.)

Question: how does your child communicate?

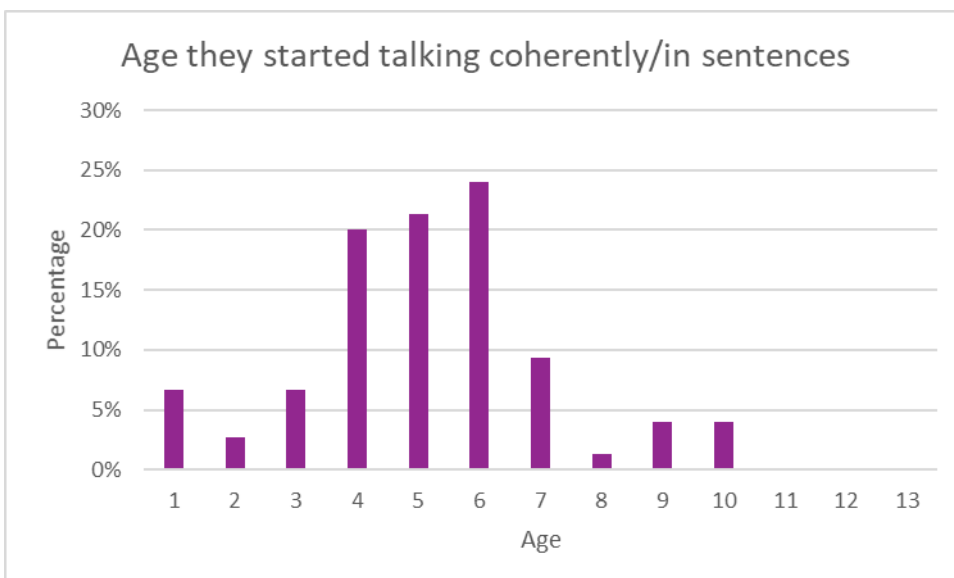
	Number	Percentage
Non-verbal	65	29%
Non-verbal – uses sign language	43	19%
Non-verbal – uses PECs	13	6%
Non-verbal – uses AAC	11	5%
Verbal – talks in 1-2 word sentences	32	14%
Verbal – talks in 3-5 word sentences	26	12%
Verbal – talks in full sentences	35	16%



### Communication – Age

44% deemed their child able to talk coherently/using sentences, and all of those were talking before 11. Just over a third of those that can talk were talking before they were 5.

Question: If your child can talk, at what age would you say they started to talk (coherently/using sentences)?



## EXTREME BEHAVIOUR

42% of children wake frequently during the night.

41% display anxiety to an excessive degree.

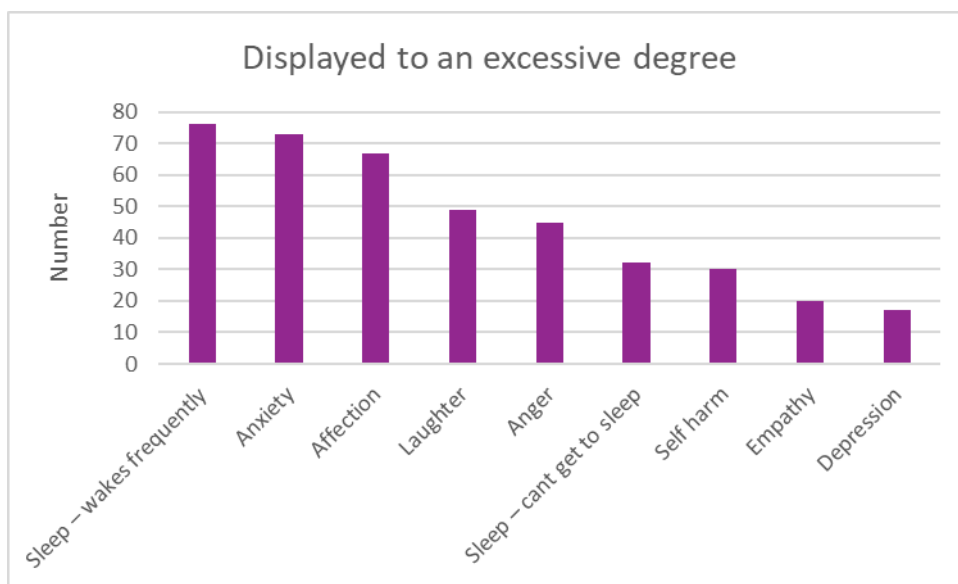
On the flip side, 37% are very affectionate and 27% laugh a lot!

17% display self-harming behaviour.

Other statistics are below.

Question: Does your child display any of the following to an excessive degree:

	Number	Percentage
Sleep – wakes frequently during the night	76	42%
Anxiety	73	41%
Affection	67	37%
Laughter	49	27%
Anger	45	25%
Sleep – can't get to sleep	32	18%
Self-harm	30	17%
Empathy	20	11%
Depression	17	9%

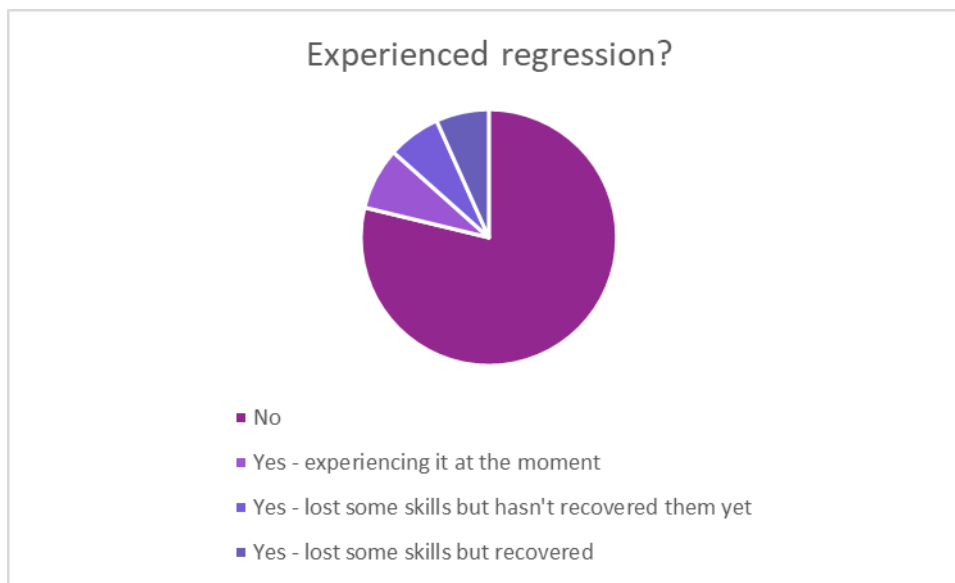


## REGRESSION

79% have not experienced regression, 14% have experienced a regression, and 8% are experiencing it at the moment.

Question: Has your child experienced severe regression? (Usually around the time of puberty/adolescence)

	Number	Percentage
No	141	79%
Yes - experiencing it at the moment	14	8%
Yes - lost some skills but hasn't recovered them yet	12	7%
Yes - lost some skills but recovered	12	7%
<i>Total</i>	179	



Regarding age groups and regression:

- 65% of all 11-20 year olds have not experienced a severe regression.
- 20% of all 21-30 year olds have not experienced a severe regression.
- 50% of all 31-40 year olds have not experienced a severe regression.

However, here we are dealing with very narrow cross sections.

# INFORMATION AND SUPPORT

This report was written by Jane du Plooy, mum to two daughters including one with Kleeftstra syndrome, and trustee of kleeftstrasynndrome.org.

Kleeftstrasynndrome.org is a small charity which exists to offer support, education and awareness of this rare condition. Over 540 families currently benefit from our community support network.

[www.kleeftstrasynndrome.org](http://www.kleeftstrasynndrome.org)

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