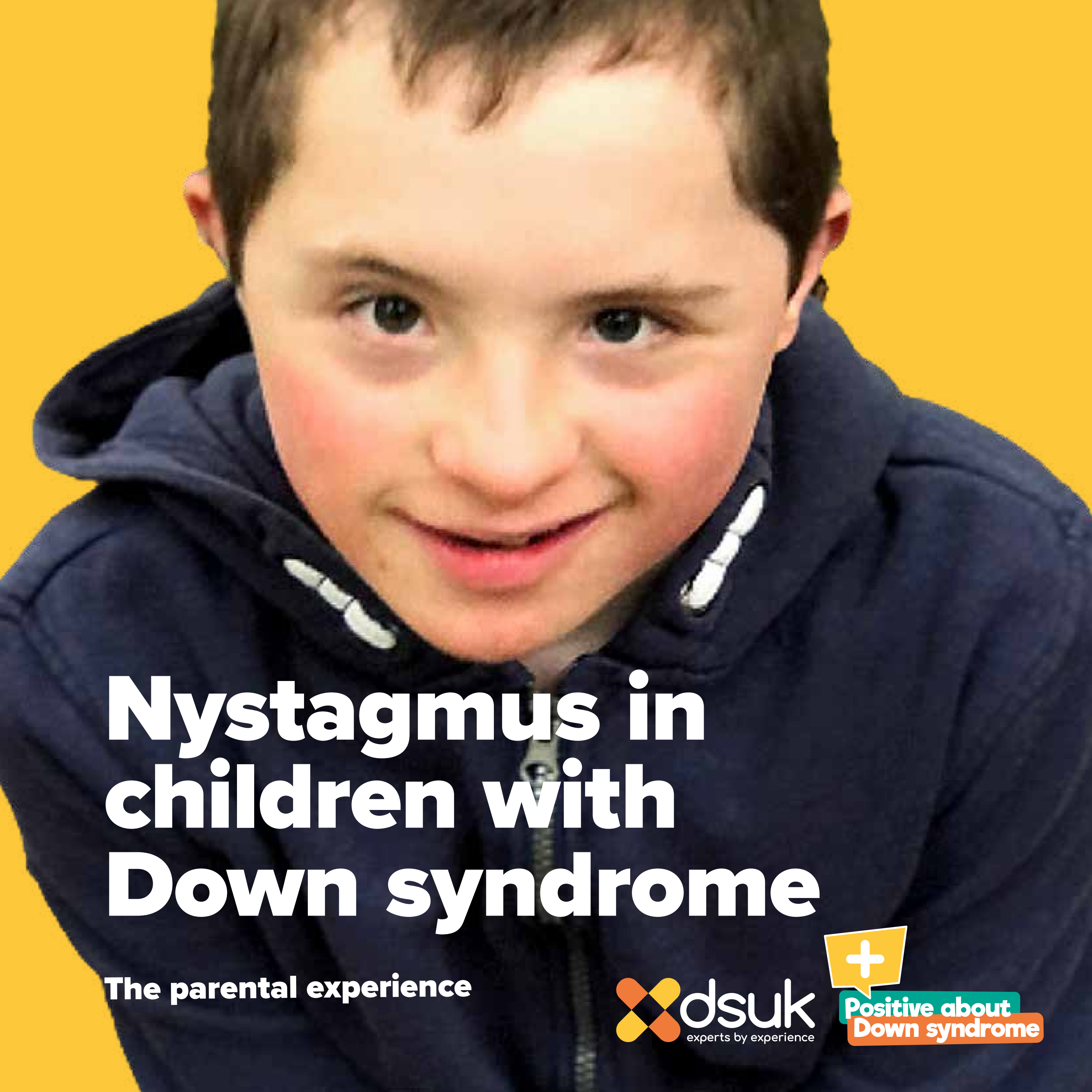


Nystagmus in children with Down syndrome

The parental experience

dsuk
experts by experience

Positive about
Down syndrome

This report is intended to raise awareness about nystagmus in children with Down syndrome, to both parents and practitioners.

The report provides an overview of the data we collected and summarises the difficulties some families and children have faced.

We are extremely grateful to Dr Maggie Woodhouse for her absolute dedication to understanding and addressing the visual needs of children with Down syndrome.

Maggie has spent almost 50 years working in the School of Optometry and Vision Sciences at Cardiff University, where she has become internationally renowned for her research into the visual requirements of children with Down syndrome. One of her most important findings is that vision is worse in children with Down syndrome than in typical children of the same age, and it remains worse even when the children have glasses – therefore, parents and schools need to be aware of that and make modifications to the learning environment. Another is that their accommodation is poor and that bifocals are of benefit to them.

Maggie approached Down Syndrome UK to collaborate on a project to explore the experiences around information and support provided to families with a child with Down syndrome and nystagmus.

Introduction

Positive about Down syndrome (PADS) works hard to ensure expectant and new parents of a baby with Down syndrome, have access to the information and support that they crave - the reality of what living with Down syndrome looks like in modern Britain, as well as ongoing peer support and expert advice when baby arrives.

We have welcomed over 200 babies with Down syndrome via our closed Facebook group Great Expectations and currently support over 120 women with a high chance/confirmed result of baby having Down syndrome. We work with approximately 1,500 families of a little one with Down syndrome under the age of 3, so are very immersed in early care for our families.

We collaborate with maternity units and universities to provide resources and training to midwives and other health professionals, with our training drawing upon the real-life lived experiences of our members together with evidence-based research.

We are passionate about ensuring parents have access to contemporary, relevant, and accurate information as well as positive and meaningful support. To this end we collaborate with several experts in their chosen field to disseminate best practice and general information, including Dr Maggie Woodhouse who is a member of PADS support team, providing information and advice to our families. Through our various networks, we also work with the families of older children with Down syndrome through to adulthood.

Nystagmus

Nystagmus is a form of visual impairment, characterised by wobbling or flickering of the eyes. It affects the ability to focus, to see the world in three dimensions and to recognise faces.

Source Nystagmus Network

There are two main types of nystagmus, one which appears in the first weeks or months of life and is called Congenital or Infantile Nystagmus and another which develops later in life which is called Acquired Nystagmus.

In this research, we contacted families with a child or young person with Down syndrome with Congenital or Infantile Nystagmus.

In the typical population, there is an estimated incidence of at least **1:1,000, c0.01%**.

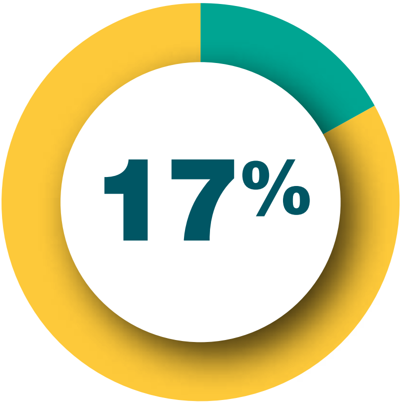
The condition is more common in children with Down syndrome. It is estimated that up to **10-15%** of children with Down syndrome have nystagmus, which equates to a population of approximately **4,000-6,000** people with Down syndrome in the UK.

Nystagmus can be a life-long visual impairment.

Key findings

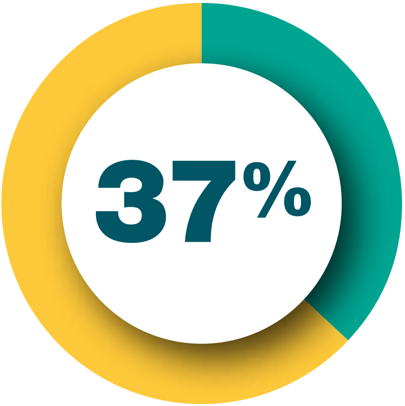
Too many children with Down syndrome and nystagmus are not receiving the care and support they deserve.

Late diagnosis



17% of children received a late diagnosis (post 2 years).

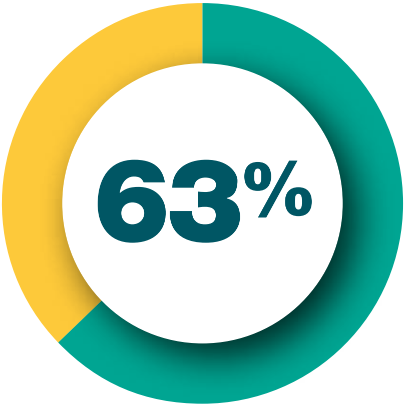
Hospitals not advising parents about the impact of nystagmus on their child's vision resulting in lack of knowledge and necessary support.



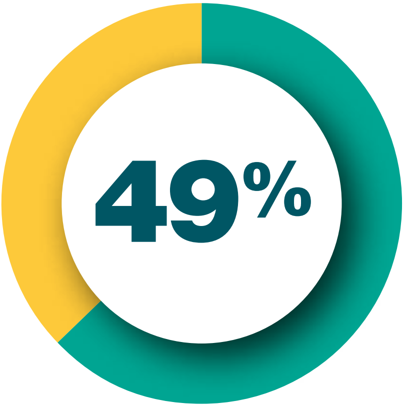
37% of families were not advised by hospital staff how nystagmus affects a child's vision.

Of these families, **70%** did not receive information from any other source.

Hospitals not referring children to Visually Impaired Services indicating lack of understanding around necessary pathway of support.



63% of children not referred to VI by the hospital.



49% of children not referred to VI at all.

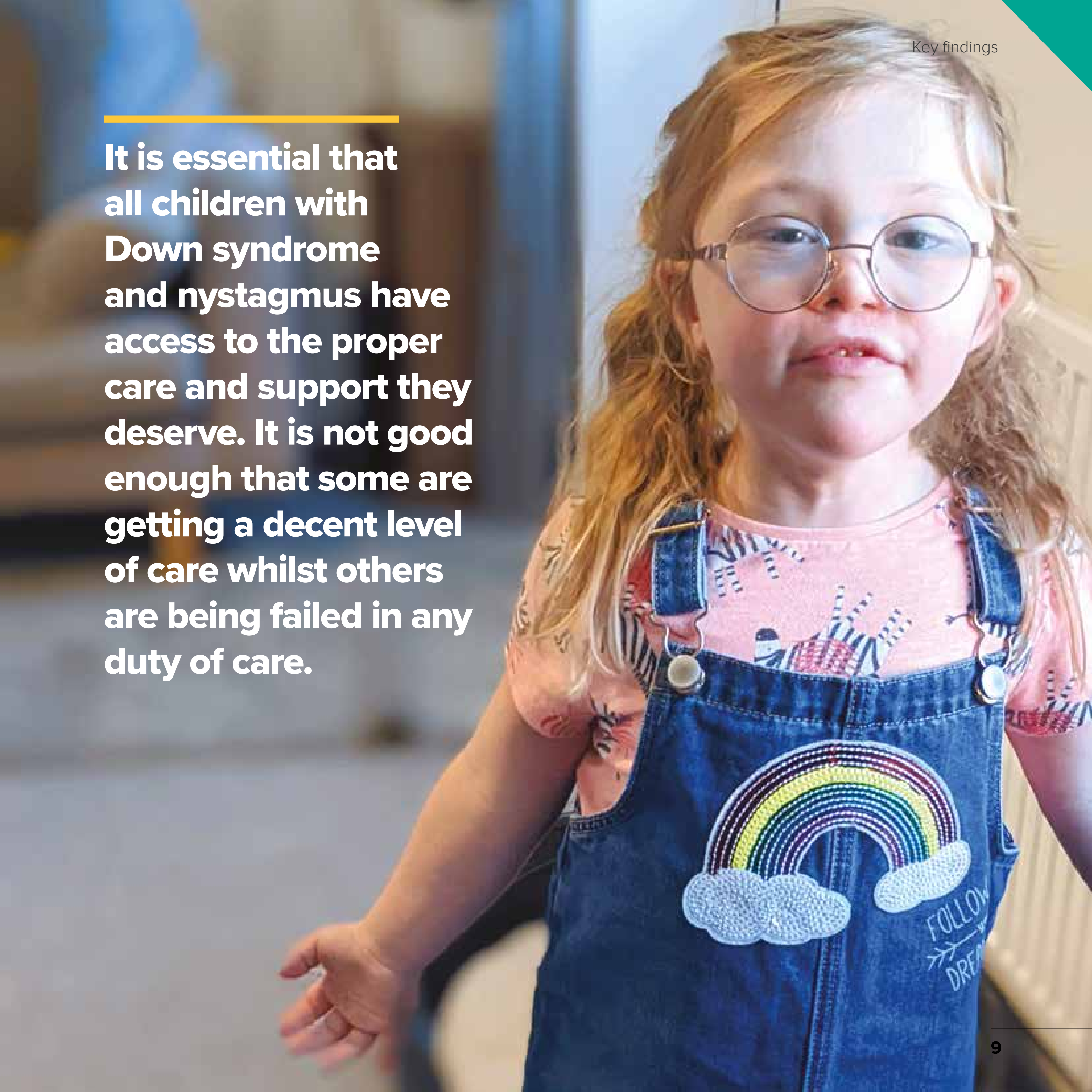
Key findings

Having nystagmus confers several visual disadvantages on children. Through organisations such as Nystagmus Network, there is information and practical advice around supporting children with nystagmus. However, if parents are not informed that their child has additional visual needs, they do not know to seek such information and support, and the child does not receive the specialist intervention around their vision that they need.

Our survey showed that parents are not always aware that having nystagmus impacts their child with Down syndrome's vision. We discovered the somewhat shocking statistic that one third of parents were not told anything by the hospital, about how nystagmus affects their child's vision.

Parents are all too often left to find information for themselves, or in some cases, parents never realise that their child's vision is poor. In such cases, children's difficulties are probably attributed to their learning disability, when simple adjustments at home and school could enable the child to learn far more easily.

The data presented from the survey suggests that there is a lack of provision and understanding for children with Down syndrome and nystagmus. Children have not been diagnosed early enough, those who have been diagnosed haven't been referred to the right specialists, parents have been misguided by hospitals and there is a severe lack of guidance for parents who have children with both Down syndrome and nystagmus.



It is essential that all children with Down syndrome and nystagmus have access to the proper care and support they deserve. It is not good enough that some are getting a decent level of care whilst others are being failed in any duty of care.

Feedback

My child was only referred to VI, as I asked because Maggie recommended it. I was just handed a leaflet and told he can see from certain parts of his eye.

It was only when we went to Cardiff to see Maggie that she explained our son has Nystagmus and what it was. He was under our local hospital for 3 years and they never gave us a diagnosis.

My son controls his nystagmus and has developed coping strategies, because of this the doctor stated they won't provide glasses.

It would be nice to know how nystagmus affects my son but his doctor said they won't know till he is 5 or 6.

Despite having nystagmus our daughter is no longer supported by VI Specialist Teaching Service, but school do put in interventions such as yellow lines paper, larger fonts etc.

It feels like something else we have to deal with alone. There's such a lack of information and support from the NHS, thank goodness for Maggie and PADS!

Visual acuity

How children with Down syndrome with nystagmus see, compared to typical children with nystagmus

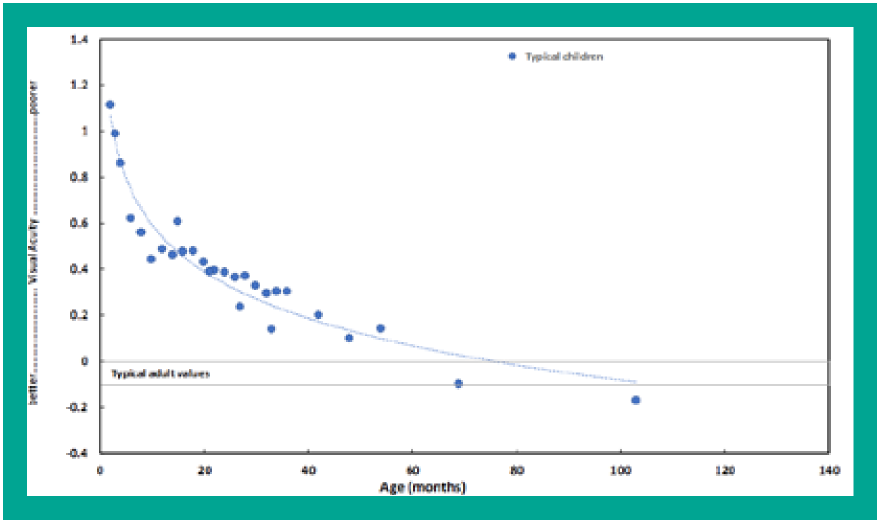
Visual acuity refers to a person's ability to see small details. It is also referred to as clarity of vision or sharpness of vision.

A visual acuity score is the result of a vision test performed by an optometrist or ophthalmologist. Opticians, technicians, and nurses can also perform a visual acuity measurement.

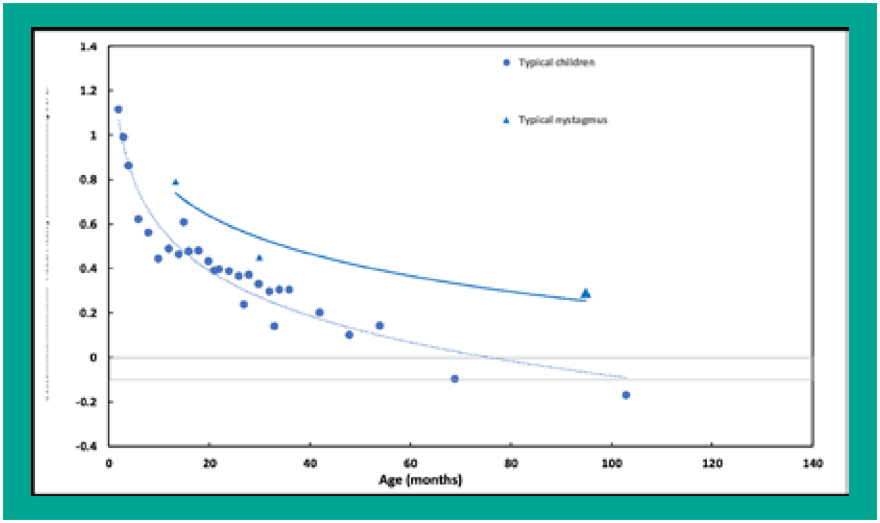
The data from the following graphs emphatically and clearly shows that children with Down syndrome and nystagmus have significantly poorer vision than that of a child with just nystagmus.

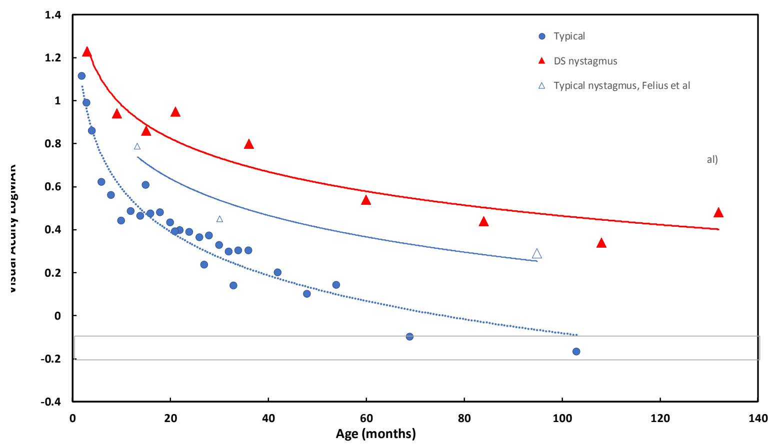
Source: www.visioncenter.org/resources/visual-acuity-score/

Visual Acuity (detail vision) – typically developing children

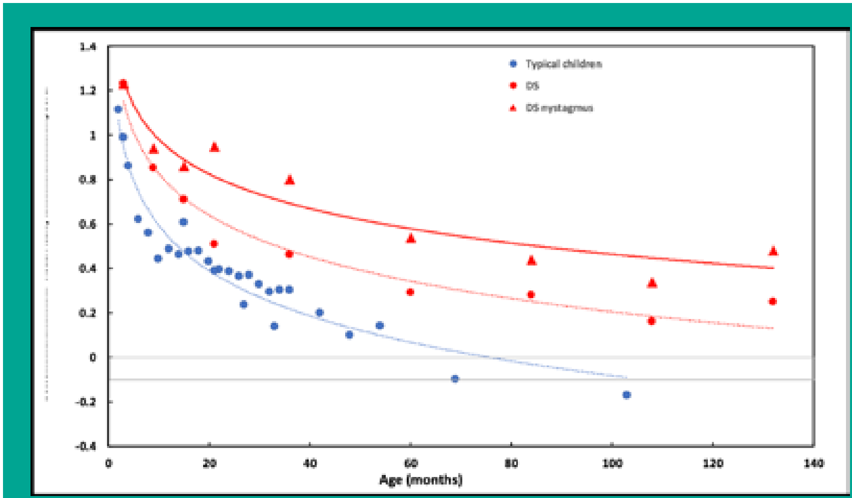


Visual Acuity – typically developing children with nystagmus





Visual Acuity – typically developing children with nystagmus



About the survey

Down Syndrome UK, in collaboration with Dr Maggie Woodhouse, Senior Lecturer and Head of the Down's Syndrome Visual Unit at Cardiff University, conducted a survey in December 2020.

A total of 59 respondents were recruited online, with the parents of 55 children with Down syndrome and a confirmed diagnosis of nystagmus responding, together with the parents of a further 4 children awaiting diagnosis for nystagmus. Families represented regions from across the country, and the data highlights that the lack of services relating to nystagmus in children with Down syndrome is a nationwide issue.

Questions in the survey

1. Has your child been diagnosed with nystagmus?
2. Which region of the UK are you based in?
3. At what age was your child diagnosed, or at what age do you expect them to be diagnosed, with nystagmus?
4. Who diagnosed your child as having nystagmus?
5. Some children grow out of having nystagmus. If your child has, at what age did this occur?
6. How long did your child have the condition?
7. What is your child's age now?
8. Has it ever been explained to you by hospital staff how nystagmus affects the way your child sees?
9. Has anyone outside of the hospital clinic given you an explanation of how nystagmus affects the way your child sees? If so, please advise who eg, Down syndrome charity, optometrist/optician at local optician's, teacher etc.
10. Have you been referred to a VI service (teacher of the Visually Impaired)?
11. Who referred you to Visually Impaired services?
12. Many thanks for completing this survey. If you wish to share any other information on this subject, please do so!

Exploring the data

QUESTION 1

HAS YOUR CHILD BEEN DIAGNOSED WITH NYSTAGMUS?

Of the **59 parents** who answered the survey, **55** have children who have been diagnosed with nystagmus, with **4** who were awaiting a diagnosis.

QUESTION 2

WHICH REGION OF THE UK ARE YOU BASED IN?

Whilst the largest cohort of respondents **(39%)** are based in the South of England, families from across the UK participated.

Scotland	**6.78%**	**4**
North West	**11.86%**	**7**
North East	**11.86%**	**7**
West Midlands	**10.17%**	**6**
East Midlands	**15.25%**	**9**
Wales	**1.69%**	**1**
South West	**13.56%**	**8**
South East	**25.42%**	**15**
Northern Ireland	**3.39%**	**2**

QUESTION 3

AT WHAT AGE WAS YOUR CHILD DIAGNOSED, OR AT WHAT AGE DO YOU EXPECT THEM TO BE DIAGNOSED, WITH NYSTAGMUS?

Question 3 suggests too many children with Down syndrome and nystagmus are being left undiagnosed for too long. The majority of children are being diagnosed by 6 months of age (42%), and one would expect a diagnosis by the age of 24 months. However, 17% of the children were diagnosed above this threshold. The data provides evidence of health services letting some children with Down syndrome and nystagmus down. It is essential that children with Down syndrome and nystagmus are diagnosed as soon as possible to give parents the necessary information and guidance needed to support their child's development.

0-6 months	**42.37%**	**25**
6-12 months	**22.03%**	**13**
12-18 months	**15.25%**	**9**
18-24 months	**3.39%**	**2**
2-3 years	**8.47%**	**5**
3-4 years	**3.39%**	**2**
4-5 years	**0.00%**	**0**
5+ years	**5.08%**	**3**

QUESTION 4

WHO DIAGNOSED YOUR CHILD AS HAVING NYSTAGMUS?

As one would expect, the overwhelming majority of children were diagnosed by ophthalmologists, although some parents mention paediatricians, one assumes and hopes the family were referred on to eye care specialists.

QUESTION 5

SOME CHILDREN GROW OUT OF HAVING NYSTAGMUS. IF YOUR CHILD HAS, AT WHAT AGE DID THIS OCCUR?

This data proves that it's essential that children with Down syndrome and nystagmus are diagnosed early, as the condition - in most cases - lasts well into childhood, if not life.

0-6 months	**6.78%**	**4**
6-12 months	**0.00%**	**0**
12-18 months	**1.69%**	**1**
18-24 months	**3.39%**	**2**
2-3 years	**3.39%**	**2**
3-4 years	**1.69%**	**1**
4-5 years	**0.00%**	**0**
5+ years	**0.00%**	**0**
Not applicable - still has nystagmus	**77.97%**	**46**
It is now only visible when one eye is covered	**5.08%**	**3**

QUESTION 6

HOW LONG DID YOUR CHILD HAVE THE CONDITION?

Less than a year	**3.39%**	**2**
1 year	**3.39%**	**2**
2 years	**3.39%**	**2**
3 years	**5.08%**	**3**
5 years	**0.00%**	**0**
6 years	**0.00%**	**0**
7 years	**0.00%**	**0**
8 years	**0.00%**	**0**
9 years	**0.00%**	**0**
10 years +	**0.00%**	**50**
Ongoing, still has nystagmus	**84.75%**	**59**

QUESTION 7

WHAT IS YOUR CHILD'S AGE NOW?

The mean age of the children in this survey, excluding those over ten years old (and making under 1 year 0), is 3.2. Over half of these children are three years old or younger. In relation to question 3, the data from question 7 outlines that the problem of late diagnosis isn't outdated but is in fact an issue that's current.

Under 1 year	**10.17%**	**6**
1 year	**8.47%**	**5**
2 years	**18.64%**	**11**
3 years	**18.64%**	**11**
4 years	**0.00%**	**0**
5 years	**3.39%**	**2**
6 years	**1.69%**	**1**
7 years	**3.39%**	**2**
8 years	**5.08%**	**3**
9 years	**5.08%**	**3**
10 years +	**25.42%**	**15**

QUESTION 8

HAS IT EVER BEEN EXPLAINED TO YOU BY HOSPITAL STAFF HOW NYSTAGMUS AFFECTS THE WAY YOUR CHILD SEES?

Data from the answers to question 8 reveals the most shocking evidence of a lack of provision for children with Down syndrome and nystagmus. Over a third of parents haven't been told how nystagmus affects the way their child sees. As seen in the graphs presented, children with Down syndrome and nystagmus have severely affected vision compared to a typical child without nystagmus. It is essential that parents understand how their child's vision is affected to best accommodate their needs. In addition, it's a terrible revelation that even when diagnosed, hospital staff haven't explained to parents how their child's vision is affected. It suggests a lack of understanding on a subject which affects a vast number of people and a lack of resources, both of which are trends that can be seen by parents in various cases across the Down syndrome community.

Yes	**50.85%**	**30**
No	**37.29%**	**22**
Not sure	**11.86%**	**7**

QUESTION 9

HAS ANYONE OUTSIDE OF THE HOSPITAL CLINIC GIVEN YOU AN EXPLANATION OF HOW NYSTAGMUS AFFECTS THE WAY YOUR CHILD SEES? IF SO, PLEASE ADVISE WHO EG, DOWN SYNDROME CHARITY, OPTOMETRIST/OPTICIAN AT LOCAL OPTICIAN'S, TEACHER ETC.

It is worth noting that of 20 parents who had not received an explanation from the hospital, 70% have not received an explanation by anyone. Of those who did not receive information from the hospital, 1 parent received information by a medical colleague, 2 from charities and 3 from Maggie Woodhouse.

There clearly is not sufficient support for parents of children with Down syndrome around their child's diagnosis of nystagmus.

- Of the 54 responses, 23 of the parents said that they had not been given an explanation outside of the hospital.
- 7 parents advised that a specialist optometrist told them, either in person or through a Facebook post by PADS.
- 9 parents were given an explanation by a Visual Impairment specialist.
- 5 parents advised they did their own research online.

QUESTION 10

HAVE YOU BEEN REFERRED TO A VI SERVICE (TEACHER OF THE VISUALLY IMPAIRED)?

Data from question 10 once again outlines the significant lack of provision for children with Down syndrome and nystagmus. Although there are indeed different severities of nystagmus, it's simply not good enough that almost half of children diagnosed with nystagmus have not been referred to a Qualified Teacher of the Visually Impaired. We know that children with Down syndrome and nystagmus have more severely affected vision than children without Down syndrome and nystagmus. When there is evidence to show that nystagmus affects children with Down syndrome to a greater extent than a typical child, the fact that so many with both conditions aren't being referred to a specialist suggests that there is a lack of understanding of what provisions and help are required.

Yes	**50.85%**	**30**
No	**49.15**	**29**

QUESTION 11

WHO REFERRED YOU TO VISUALLY IMPAIRED SERVICES?

Question 11 was only answered by those who had been directed to VI services. In the ‘other’ option, specifications included: an SEN team, portage, a hearing impairment specialist, IDS (Integrated Disability Service) and someone whose child was already under Visually Impaired service due to another visual impairment.

Data from question 11 provides further evidence that hospitals are not providing parents the support that is required for children with Down syndrome and nystagmus. In corroboration with findings from question 10, the fact that nearly two-thirds of parents were not referred to Visually Impaired services by a hospital - and were subsequently referred by someone else – strongly suggests that hospitals across the country do not have sufficient understanding of what pathway of support is required for children with Down syndrome and nystagmus.

Ophthalmologist or orthoptist at the hospital	**36.67%**	**11**
Optometrist at local practice	**0.00%**	**0**
Specialist clinic at a university	**13.33%**	**4**
Paediatrician	**13.33%**	**4**
GP	**0.00%**	**0**
Health Visitor	**0.00%**	**0**
Child's nursery or school	**10.00%**	**3**
Other (please specify)	**26.67%**	**8**

QUESTION 12

MANY THANKS FOR COMPLETING THIS SURVEY. IF YOU WISH TO SHARE ANY OTHER INFORMATION ON THIS SUBJECT, PLEASE DO SO!

A key addition and important part of the survey was to allow parents to give more information on their experience. The extent of the lack of understanding and provision is highlighted by some of the additional responses.

- One parent said that they no longer get VI specialist help, but that their school is useful and gives extra provision to help their daughter.
- Another parent said that the school have been very useful, and they'd struggle with their child's mobility were it not for help from the school.
- Three parents said they'd like more information to be available on the subject to help their children.
- One parent was told that they wouldn't know how nystagmus would affect their son until he was 5 or 6.
- One parent mentioned that although their child was diagnosed between 6-12 months old, they noticed it at 8 weeks and had to wait for months to get an appointment.

These responses emphasise how unacceptable the treatment of some children with nystagmus has been and that there is an overwhelming lack of support for too many parents. Parents have been supplied with information that isn't only lacking in clarity and depth, but also information that simply isn't accurate.

Further reading

Resources recommended by parents and by Maggie Woodhouse

- Moorfields Eye Hospital
  www.moorfields.nhs.uk
- Nystagmus Network
  www.nystagmusnetwork.org
- RNIB
  www.rnib.org.uk/eye-health/eye-conditions/nystagmus
- Somerset Sight
  www.somersetsight.org.uk
- Specsavers
  www.specsavers.co.uk/eye-health/nystagmus
- Vision Support
  www.visionsupport.org.uk
- DSUK has a closed Facebook group exclusively for optometrists with an interest in working with people with Down syndrome.
  www.facebook.com/groups dsukoptometristsworkingwithpeoplewithdownsyndrome

> Thank you for undertaking this research. I don't think there is enough support or understanding of this condition for our children. I think generally people in education underestimate the impact it has on children and young people. Also lack of understanding on things like whether or not to be registered blind, the impact on independence (crossing roads, looking for bus numbers etc) Also treatments - my daughter had surgery to alter the null point to help with head posture. Hideous operation but worth it, I think. Again, felt quite alone when trying to make the decision on whether to go ahead or not.

www.downsyndromeuk.co.uk
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Positive about Down syndrome

dsuk
experts by experience

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School of Optometry
and Vision Sciences

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