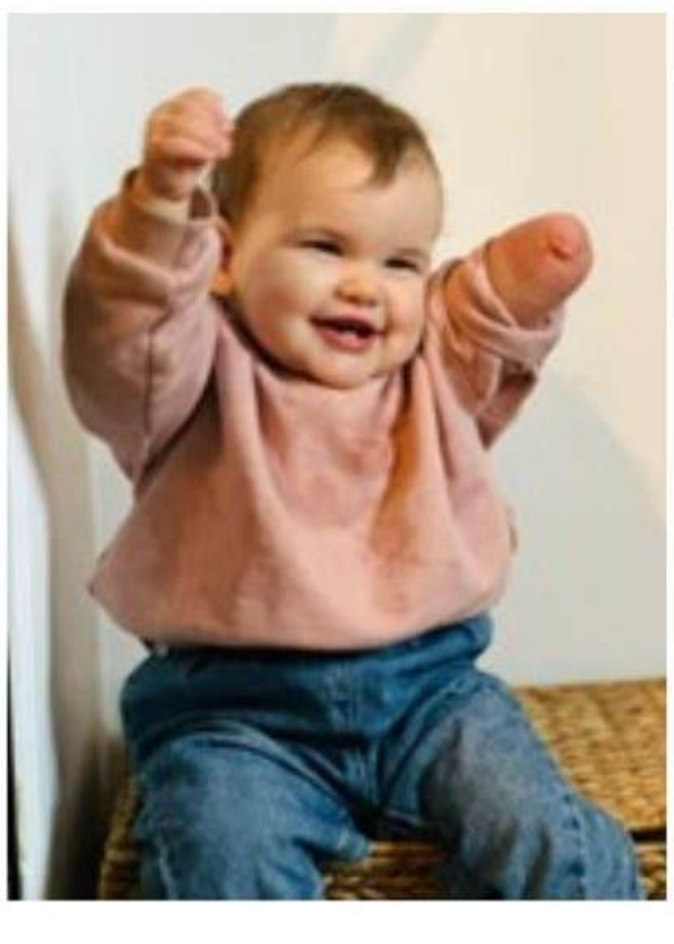
Looking beyond the Visual Difference at birth

How to support the parents of a baby with a visual difference of the upper limb

Ruth Lester OBE FRCS, Retired Consultant Plastic Surgeon and trustee for the REACH charity

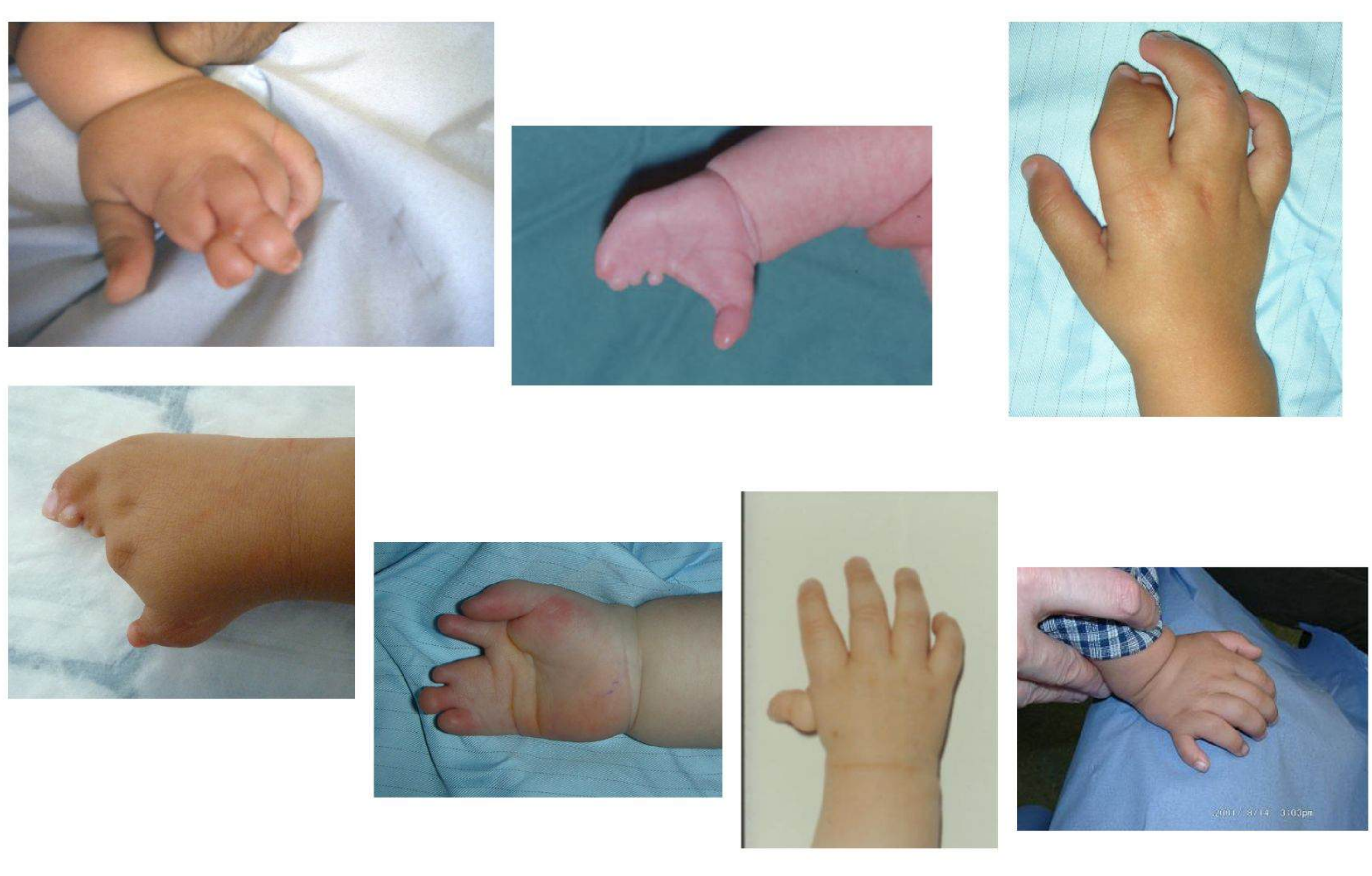
Congenital differences of the upper limb are a rare disorder, and midwives may only see a handful of babies with this condition during their professional lifetime. However, parents' experience of receiving the news that their baby has a limb difference is affected significantly by the approach of the healthcare first responders. Ruth Lester, retired Consultant Plastic Surgeon and trustee for the REACH charity, explains how midwives may help provide the support for parents using a positive approach.

(Please note a version of this article was published in RCM Midwives October 2024)



Background
Congenital Upper Limb Anomalies are amongst one of the rare conditions which affect about 19.2 to 27.2 per 10,000 live births. (Ref 1). However, the range of differences is very great from simple accessory digits to complex presentations including the elbow, forearm, wrist, and hand. Over 75% of hand differences occur in otherwise healthy babies without other non hand anomalies (Ref 2 and 3).

Antenatal Ultrasound
The ability to detect fetal anomalies has improved significantly over the last few years and an ultrasound examination at around 20 weeks of gestation is now a routine part of antenatal care. However, only around 50% of anomalies like these are picked up at the scan but hand only anomalies are much less likely to be seen via this medium (Ref 4).
These are the **types of differences** which may not be picked up antenatally.



Extract from RCOG Guidelines 2010
If the scan reveals either a suspected or confirmed abnormality, the mother should be informed by the sonographer at the time of the scan. It is essential that all practitioners performing fetal anomaly ultrasound screening should be trained to communicate abnormal findings to women, as such information is likely to have significant emotional impact. (Ref 5 and 6).

Termination of pregnancy for fetal abnormality may only be considered if there is a substantial risk that the child, if born, would suffer physical or mental abnormalities that would result in serious handicap. It is difficult to define the word 'serious' in this situation as we know that the majority of babies born with an upper limb difference will live active and fulfilled lives. (See Reach https://reach.org.uk).

A note on Terminology
Although, we, as professionals tend to use the word 'anomalies' and 'abnormal' in our discussions, it is worth bearing in mind that these words can be very emotive for the community. It is best to use the word 'Difference' wherever possible.

'Reach Charity' is an organisation which supports children and their families with upper limb differences. The charity heard from new parents in particular that they were going through a very traumatic experience of discovery of the anomaly in their baby both antenatally and at birth.

We therefore set out to evaluate the parental experience of the news that their child has a Congenital Upper Limb Difference (CULD) in the UK and Ireland.

Parental experience after diagnosis of a congenital upper limb difference: a national survey. (Ref 7)
The aims of the survey were as follows:
1. Determine when and how parents first learnt of an upper limb difference.
2. To identify what information was given.
3. To understand the parental emotional aspects of the journey to discovery of the diagnosis and cause of the CULD.
4. To ask how the journey could be improved

This was a mixed methodology survey carried out online with parents of a child under 10 years old affected by an upper limb difference from UK and Ireland. Although the main focus was from Reach, we did invite other parents from other organisations via social media to contribute

Highlights of the study
1. There were 261 responses throughout the UK and Ireland with a wide variation of conditions but around 25% of parents did not know the diagnosis of the condition of their child.
2. The upper limb difference was first suspected antenatally in 41.1%.
3. Only 28% of parents felt that they were given relevant information before they left the hospital
4. Only 54% of the families were seen by a specialist within 3 months
5. Over 90% of parents searched for information themselves on the internet

On analysing the themes, parents described the following:

1) Lack of support
'Each doctor said something different. Everyone we asked had no answer and sent us to another person. There was no support from anyone. Was just told to join lucky fin' (An American based charity)
Respondent 82, London
2) Concerns about the future for their child (including underlying cause, treatment and wider functional and life implications)
'Insights into limb function & treatment, plus whether any related conditions or wider implications'
Respondent 12, Southeast England
3) No clear communication about what would happen next
'Just that it was called an upper limb difference would have been helpful for a start! We didn't even know where to begin… how long it would be till we could see someone about it. Some acknowledgement of the hugeness of this life altering thing – the midwife didn't even notice.'
Respondent 226, Northwest England
4) Offer of a termination
'The narrative was very negative with lots of apologies'
Respondent 6, Yorkshire and the Humber

What can you do?
Midwives are on the frontline when it comes to news of a congenital upper limb difference. It is a rare event and as midwives you may only see a very few cases during your working life.

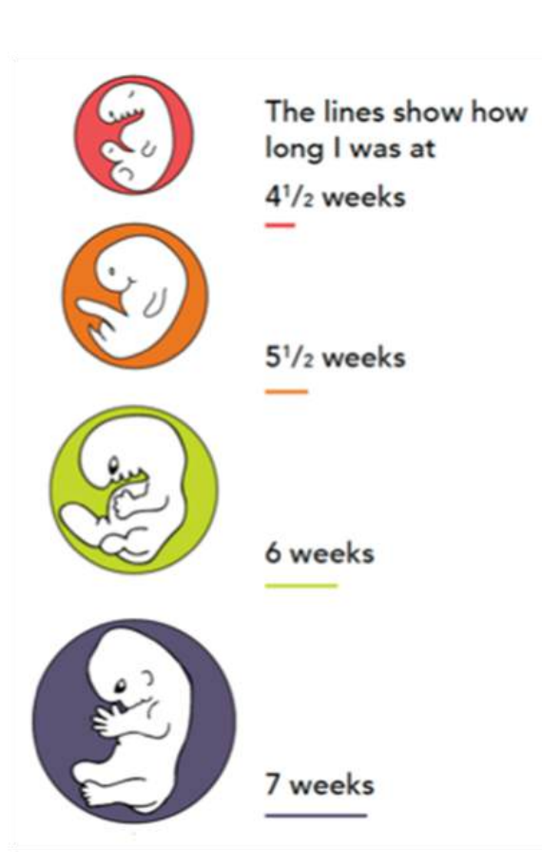
These conditions are rarely life limiting but do affect the way the child may function and feel about themselves. However, children born with upper limb differences are extremely adaptable. The positivity of front-line healthcare professionals is very helpful to parents who are experiencing shock, grief, guilt etc. when faced with a baby with a highly visible difference

Because parents are in shock, their cognitive state isn't able to retain information and or process some things easily. They need time and reassurance that the shock will settle.#

Some Do's and Don'ts
1) Don't ask questions linked to cause – e.g. family history or events in pregnancy (Parents feel guilty at this time and the questions raise doubts in their minds)
2) Don't say it's the first time you've seen it, even if it is – just say we need more information. (Parents need to feel some security)
3) Use neutral language – e.g. dysmelia, upper limb difference
4) Don't offer a solution or pretend it never happened – be realistic!
5) Termination is very controversial and the mention of it can lead to a lot of negativity by the parent
6) Do respond to questions asked

We would like to encourage you to reassure and support parents – **their child's condition was not their fault!** (Ref 7, 8, 9)

Understanding the embryology



A baby's hand is fully formed by 6-7 weeks after fertilisation – i.e. Before mum even knows she is pregnant!

The limb 'patterning' during those early few weeks is very complex and therefore not so surprising that there is sometimes a 'mistake'.

It only takes one cell to get the 'messaging' out of line for these differences to occur.

Learning points:
1. For **single limb involvement**, further consultation is needed but reassurance can be given that child will most likely **live a normal and productive life**.
2. If there is **bilateral involvement** further investigations will needed but the children are still more than likely to have a **relatively normal successful life**
3. Where **multiple anomalies** are detected, **urgent referral** is appropriate for fuller assessment. It is therefore more **difficult to offer immediate complete reassurance**. Parents need to know that they are on a journey of discovery and know who and when a specialist will see them.

How you can reassure:
1) Be honest
2) Be positive
3) Take away the guilt by understanding the embryology
4) Ensure access to a specialist as soon as possible

Refer to:

Reach

https://reach.org.uk

A network of families whose children have upper limb difference, and people with upper limb difference.

Further Reading:

1) Ekblom AG, Laurell T, Arner M. Epidemiology of congenital upper limb anomalies in 562 children born in 1997 to 2007: a total population study from stockholm, sweden. J Hand Surg Am. 2010 Nov;35(11):1742-54. doi:10.1016/j.jhsa.2010.07.007. Epub 2010 Oct 20. PMID: 20961708.Wildenauer

2) L, Mavrommatis S, Bae DS, Steinman SE, Wall LB; CoULD Study Group; Van Heest AE. Syndromic Involvement of Patients Presenting With Congenital Upper Limb Anomalies: An Analysis of 4,317 Cases. J Hand Surg Am. 2024 Apr;49(4):311-320. doi: 10.1016/j.jhsa.2023.11.020. Epub 2024 Jan 14. PMID: 38231172.

3) Piper SL, Dicke JM, Wall LB, Shen TS, Goldfarb CA. Prenatal Detection of Upper Limb Differences With Obstetric Ultrasound. J Hand Surg Am. 2015 Jul;40(7):1310-1317.e3. doi: 10.1016/j.jhsa.2015.04.013. Epub 2015 May 28. PMID: 26026354; PMCID: PMC4568827.

4) RCOG (2010) Termination of Pregnancy for Fetal Abnormality in England, Scotland and Wales. Available at: https://www.rcog.org.uk (Accessed 22.3.2021)

5) Johnson J, Arezina J, Tomlin L, et al. UK consensus guidelines for the delivery of unexpected news in obstetric ultrasound: The ASCKS framework. Ultrasound. 2020;28(4):235-245. doi:10.1177/1742271X20935911

6) Clelland AD, Lester R, Duncan Ó, Lam WL. Parental experience after diagnosis of a congenital upper limb difference: a national survey. J Hand Surg Eur Vol. 2024 May 3:17531934241249014. doi: 10.1177/17531934241249014. Epub ahead of print. PMID: 38702055.

**7) Managing broken expectations after a diagnosis of fetal anomaly
Rosemary Theroux, Cheryl L. Hersperger SSM – Qualitative Research in Health 2 (2022) 100188**

**8) Parent Experiences and Preferences When Dysmelia Is Identified During the Prenatal and Perinatal Periods: A Qualitative Study into Family Nursing Care for Rare Diseases
Judith Johnson, PhD, ClinPsyD1,2 , Olivia Johnson, BSc(Hons)3, Jane Heyhoe, PhD2, Charlotte Fielder, MBE4,5, and Alice Dunning, BSc(Hons). Journal of Family Nursing 2018, Vol. 24(2) 271–293**

**9) Congenital upper limb anomalies: Ruth Lester and Professor Mark Kilby on what to say to parents if such an anomaly is found at the 20-week antenatal scan
Synergy News July 2021 ISSN 1741-4245**

**10) Looking beyond the Hand Difference MM Hour Series 13 no 10
https://www.youtube.com/watch?v=D_1m9U2rqQM**

Reach

'Reach Charity' is an organisation which supports children and their families with upper limb differences and can be contacted at: https://reach.org.uk

Ruth Lester OBE FRCS
Retired Consultant Plastic Surgeon and trustee for the REACH charity
January 2025