

Not quite perfect! Diagnosis of a minor congenital abnormality during examination of the newborn

Due to advances in antenatal screening programmes, parents may have unrealistic expectations that their child will be born 'perfect'. The examination of the newborn can shatter this illusion when minor congenital anomalies, not detected by antenatal screening, are diagnosed. This paper endeavours to explore parental reaction when the diagnosis of polydactyly and/or syndactyly in the term infant is made and discuss how staff can facilitate the parents in accepting their 'not quite perfect' child.

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Key points

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1. Parents may have unrealistic expectations that their child will be born 'perfect' after negative antenatal screening.
2. The practitioner's knowledge of cause, prognosis and treatment of common congenital abnormalities is paramount if appropriate support is to be given.
3. An understanding of bereavement and parental reactions to congenital abnormalities will enable the examiner to facilitate the parents in accepting their 'not quite perfect' child.

Foreword

This piece of work was initially done as part of staff training to carry out the examination of the newborn. It was prompted by two observations. The first was the birth of a baby with polydactyly, in this case an extra finger on one hand, which was attached by a small piece of skin. The parents were absolutely devastated that their baby wasn't perfect. They insisted that throughout the pregnancy they had done everything they had been told to do. They had complied with all the screening programmes and scans, which they had been told, were all 'normal' and did not understand why this anomaly had not been detected. They immediately blamed the medical staff for not detecting the birth defect.

Although the parents were told that because the extra finger was non-functioning and only attached by a small skin tag it could be removed surgically at a later date, with minimal scarring, they remained totally fixated upon it, seemingly unable to accept that their baby was otherwise perfect. They wanted answers, which obviously could not be given, and blamed the system for their 'not quite perfect' child.

The second observation was a baby born with syndactyly of both feet. The parents were quite accepting of this despite the fact it had also not been picked up on scan. Syndactyly was a familial trait and they felt that the anomaly made their baby as unique as other members of their

immediate family.

These two differing reactions to a birth defect affected me deeply. I was embarking on a training programme whereby I may be faced with informing parents that their baby had a birth anomaly and these experiences made me concerned as to how I would handle the situation and how the parents would react. The following paper attempts to assist the practitioner's management of such a situation.

Literature review

Before embarking on this project a thorough literature review of the available articles around parental reaction to birth anomalies was conducted. This revealed that although there is evidence of research examining their reactions to an identified birth defect, there is relatively little available regarding the psychological effects on parents of a child with an unidentified birth anomaly, especially after false negative results. There was only one paper by Hall et al¹ which discussed false negative results on prenatal screening for Down's syndrome. This appears to be the first systematic attempt to document the parental response to a false negative result. They reported no differences in levels of anxiety and depression but noted that the mothers in the false negative group had higher parenting stress and more negative attitudes towards their children. Both mothers and fathers were more likely to blame others for the outcome, particularly health professionals or the medical system

in general for not detecting the affected child prenatally.

Another exploratory study using qualitative methods reported the main outcome variable as being the mothers' expression of suspended mothering: a diminished maternal role, a feeling of being uninvolved in their baby's future². There is also evidence that fathers felt that they were often not included in discussions and that the focus was mainly on the mother and child³.

The available literature also suggests that despite the National Screening Committee's attempts to advise that antenatal screening programmes are based on 'risk reduction'⁴, parental perception of the screening tests still remains ambiguous and there remains a misconception that negative/normal means 'perfect'^{1,5}. This ambiguity appears to be confirmed in an article published by Maia in which the author – a parent – discusses, in-depth, the limitations and risks of false negatives of the AFP (alpha-fetoprotein) screening process for Down's syndrome and the lack of information given to parents at the time of the test⁶. However, although she obviously understands 'risk reduction' she states "Thank heavens, in my case, the AFP test was negative and I didn't give it another thought!" This statement is surprising given all the information she had, that she still saw the AFP screening programme as a 'guarantee' that she would have a 'perfect baby'.

Is it any wonder then that many parents are confused and unable to comprehend that their child has a birth anomaly when they have 'done all the right things' and, as far as they are concerned, being advised that a screening process is 'negative' or 'normal,' reinforces their belief that their baby is 'perfect'⁵.

Something's wrong!

It is during the examination of the newborn that the diagnosis of polydactyly or syndactyly will be made. Polydactyly is defined as extra fingers or toes (**FIGURE 1**), and syndactyly (**FIGURE 2**) is the webbing of hands or feet.

Although historically the examination is performed by medical staff, other professionals such as advanced neonatal nurse practitioners, neonatal nurses and midwives are now taking on this extended role. It follows, therefore, that an element of this role would be to inform parents that their baby has a congenital abnormality.



FIGURE 1 An example of polydactyly. Photo: Science Photo Library.



FIGURE 2 An example of syndactyly showing webbing of the hand.

The initial examination, by the midwife, is generally brief, and will normally exclude any major anomalies. As it may be some time before the baby has its comprehensive newborn examination, the parents may have meticulously examined their own child, and are usually the first to identify abnormalities and, therefore, alert staff to a problem before the examination⁷.

However, unless the polydactyly and syndactyly is immediately noticeable, it can be overlooked. Parents may see what they want to see, ie hands and feet with five fingers and toes, and it is not until the digits are physically separated and counted by the practitioner that the diagnosis of syndactyly or polydactyly may be made.

The parents will monitor the examination of their baby closely, and they may be acutely aware of non-verbal communication such as body language and/or silences picked up at this time, therefore, staff should be aware of their own body language during the examination. Mothers have expressed concern that the midwife did not speak directly to them⁵, and others have observed

that "whatever feelings the nurse may experience (and try to hide), non-verbal communication systems will probably convey the information anyway"⁸.

Following the findings of the examination of the newborn the parents need to be informed that their child is 'not quite perfect'. As other members of the healthcare team are now involved in this process, they need to have the necessary skills/knowledge to be able to inform and support the parents. The new *Newborn and Infant Physical Examination: Standards and Competences* guidelines regrettably give very little guidance to staff stating only that they should have knowledge of the impact of breaking bad news⁹. Given that it is well documented that divulging bad news is generally mismanaged¹⁰; the author questions whether adequate training has been included within the extended role to enable the practitioner to inform parents their baby has a birth anomaly.

Staff should, at this time, be able to provide the appropriate information and clear explanations of the anomaly, including treatments, probable/possible outcomes, support groups etc. A detailed knowledge of normal and abnormal embryonic development is essential as parents may suppose that the anomaly was caused by something they did during the pregnancy. Mothers in particular, may resort to self-blame, searching back over the pregnancy to find a cause or reason however trivial⁸. They will be anxious about their child's future and staff should be able to reassure them that support is available both within the hospital and in the community setting.

As polydactyly and syndactyly are birth anomalies not necessarily detected by ultrasound scanning, a comprehensive knowledge of false positive/negative results and limitations of antenatal screening programmes is also essential, as this may be the case for other minor anomalies¹¹.

We did everything right!

It is at this time, when faced with a baby with an anomaly that the parents may question the role of the screening processes⁴. Throughout an uncomplicated pregnancy, parents anticipate having a 'perfect' child^{5,6}. This anticipation may have been reinforced by the 'negative' or 'normal' results from antenatal screening. One parent whose daughter was born with spinal bifida states "we were not expecting our first child to be born in

anything other than perfect health; I had done all the right things²⁵.

It is also recommended that parents be given clear written advice before scanning, and this should include the limitations of the scan and the detection rates for abnormality¹¹. However, ambiguity remains “though women were knowledgeable about practical aspects of undergoing the test, they were less informed and prepared for possible adverse outcomes”, as Smith et al found¹².

There is also anecdotal evidence to question the accuracy and adequacy of the information given to parents. It is reported that parents had been “told erroneously that there could be nothing wrong with their baby²⁶”. This implies that information given at the time of screening needed to be more accurate and communicated more effectively, thus reducing any unrealistic expectations about screening programmes. The National Screening Committee state that “Screening can reduce the risk of developing a condition but cannot offer a guarantee.” There is also an irreducible minimum of false negative/positive results – screening is increasingly being presented as risk reduction, not a foolproof process⁴.

If, therefore, clear explanations relating to these limitations are not given at the 20 week anomaly scan, it is highly probable that when a birth defect such as polydactyly or syndactyly is diagnosed after the birth, parents who have had a ‘negative/normal’ scan may perceive this as a medical error and look for someone to blame^{1,5,8}.

Is this a failure of the screening programme? The anecdotal evidence appears to support the actuality that information is not always communicated accurately, thus creating confusion over the interpretation of the results.

At the same time medical advances and the advent of two, three and four dimensional ultrasonography, now provide parents with images that are more recognisable, and there is some evidence that this enhances parental-fetal bonding. Although bonding itself is a naturally occurring event that begins in the prenatal period, it still remains a complex phenomenon of which we only have a little understanding¹³. Parents may use these medical advances to build up a more detailed picture of their forthcoming child and the unidentified anomaly may have a detrimental effect on the bonding process as parents come to terms with it.

Shattered dreams!

After the initial shock of discovering their child has been diagnosed with having a birth defect, parents may go through a period of mourning for the ‘fantasy’ child they thought they were having. This is the image of the child they have built up in their own mind during the pregnancy, an image reinforced by advanced ultrasonography techniques.

Solnit and Stark were instrumental in conceptualising parental reactions to the birth of a baby with a congenital abnormality and they likened the crisis of the birth of a child with a malformation, to the emotional crisis following the death of a child. The mother must mourn the loss of her expected, normal infant, and in addition to this, she must become attached to the actual, living, but ‘not quite perfect’ child she has¹⁴. Elisabeth Kübler-Ross describes, in five discrete stages, a process by which people deal with grief and tragedy¹⁵:

- Denial
- Anger
- Bargaining
- Depression
- Acceptance

However, the mourning or grief associated with a malformed child differs because of the complex issues raised by the continuation of the child’s life, and the demands of the child’s physical care¹⁶. A hypothetical model of a normal sequence of parental reactions to the birth of a malformed child is¹⁷:

- Shock
- Denial
- Sadness and anger
- Equilibrium
- Reorganisation

Although it is probable that most parents will experience some of these feelings at some stage, each parent may well move through the complexities of mourning at a different rate⁸, “grief does not necessarily follow a neat sequence and regression to earlier stages is common, implying that it is a very individual process²⁸”. Grief can also affect the parents’ ability to communicate with each other and they should be allowed time together, to support, cry, and talk to each other, to ask questions as they come to terms with the anomaly. It was also noted that “the shock and denial reported by many parents seems to be an understandable attempt to escape the traumatic news of the baby’s malformation, so different than their

expectations for a normal healthy newborn²⁶”.

Armed with this knowledge the practitioner should also be prepared for the reality that the parents may look to someone to blame. In ancient times the bearer of the news that a battle had been lost was often killed, hence the phrase ‘kill the messenger’ which may assist staff in accepting that in the circumstances the parents may well blame them. They are identified as the official authority, the bearer of bad news, therefore they become the target to blame¹⁹.

Being there

At this time the parents may well have problems with adjusting to the reality that their child has a birth anomaly, and show signs of negativity towards the child: bad news can change a person’s view of the future in a negative way¹⁰. The parental ability to hear and process information is also affected by their emotional stress at this time, and short and concise explanations should be given²⁰. Alternative methods of imparting information around the anomaly such as leaflets, booklets, and the internet²¹ should be employed. In addition parents can receive much needed emotional, social and practical support from other parents in similar situations and information about appropriate local parent support organisations or contacts should be supplied²².

Importantly, at this emotional time, the parents need to be encouraged to touch, care for and handle their baby, and it should be pointed out to them how normal the infant is in all other respects. They should see their baby as an infant with a defect rather than a defective infant²³. Professionals should also be aware that “a family’s interpretation of the meaning of disability cannot help but reflect to some degree the larger context of social attitudes and historical realities within which that interpretation emerges.” In other words, the parental perception of the disability, however small, is based on previous experiences and how the parents may view the acceptance of their child into the wider community²⁴. Encouraging the parents to speak about how they see the anomaly affecting their child may assist with acceptance. Parents appreciated professionals giving them the opportunity to talk and show their feelings. Interestingly they also wanted the professional to show their own feelings and make a

greater effort to comfort them²⁵.

Parents may also have feelings of being disempowered and uninvolved, with no control over their baby's fate – 'suspended mothering', and it is recommended that staff facilitate maternal involvement in decision making by ensuring they are given unbiased information in a supportive manner².

It is important for the practitioner to realise that at this time the father's role can become obscured. The father will have the unenviable task of informing relatives and friends of the anomaly: at the same time they have to support their partner and deal with their own grief, something they cannot be expected to do if they are not included in decision making and care of their baby. In one study although a small sample, fathers reported that support seemed to focus on the mother and baby: also they recalled their frustration at not being included in discussions about the future of their baby⁴. It has also been suggested that fathers may feel that the anomaly is a reflection on their manhood, they may think they are incapable of producing a perfect child, causing them even more anxiety²⁶. By being aware of the potential for the father to feel excluded the practitioner can ensure that the fathers are included in all the decision making, discussions and choices around their baby and their future care.

Where do we go from here?

The changing role of advanced neonatal nurse practitioners, neonatal nurses and midwives who are now taking on the role of examining the newborn is a tremendous responsibility for the professional involved. It places them at the forefront of potentially, extremely emotional and traumatic events and it is imperative that they have adequate training and knowledge to enable them to manage these rare but probable situations.

Training around 'giving bad news' and 'grief responses' is vital so that parents can be facilitated by the practitioner to accept their 'not quite perfect' baby. An insight into the parental expectations, varying parental reactions and the stress caused to the parents, will also aid staff to offer optimum support and reassurance when informing parents of an anomaly.

To do this they need to be able to establish a relationship with the parents based on trust and truthfulness, using effective communication to give them

accurate information about the anomaly and involve them in discourse about their baby's future care. It is important to allow parents time to come to terms with the anomaly, encouraging emotional responses and reassuring them that despite 'doing everything right' these anomalies can occur at random. Staff also need to be aware of the available resources and parents know that the support they get in hospital is continued into the community.

Although there may never be a 'foolproof' system of antenatal screening which will guarantee a 'perfect' child, this paper reinforces the author's conviction that there needs to be more work done around the perceptions of the antenatal screening processes.

Perhaps it is time to move away from using words such as 'negative' or 'normal' when reporting on screening results, which may be reinforcing parental perceptions of a 'perfect' child. Rather we should be stressing the limitations of national screening programmes so that parents have a more objective view of the results. Do parents really know the implication of a 'risk factor'; is this adequately explained to them when they agree to the neonatal screening processes? The anecdotal evidence to date seems to demonstrate this is not the case. In the words of one mother "I was not given anything like enough information to make an informed choice about whether or not to have the AFP test. I was not told, and it never occurred to me to ask, because the test was presented as just one more standard, routine, everyday check that all women go through. It was never suggested that I or anyone else might ever have any reason to do anything other than tick the consent box. I ticked the box"²⁶.

If more effective communication is used it may help parents to prepare themselves more adequately for some of those anomalies which are not detected by the present antenatal screening processes. A fuller understanding of the limitations and false negatives should assist parents in accepting that 'the perfect baby' is not a guarantee, despite all the scans, tests and screening offered to them.

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