Going down a different road: first support and information needs of families with a baby with Down syndrome

Evelyne E Muggli, Veronica R Collins and Catherine Marraffa

ABSTRACT

Objective: To explore the experiences of families with a baby with Down syndrome at the time of diagnosis, and their preferences for information and support in the early period after diagnosis.

Design, setting and participants: A qualitative, interview-based study of 18 families living in Victoria with a child with Down syndrome born between 2002 and 2004 who had not been diagnosed with the syndrome before birth. Interviews were transcribed verbatim and interpretive content analysis was undertaken.

Results: Parental coping with the unexpected diagnosis of Down syndrome in their infant was influenced by the time interval between birth and disclosure of clinical suspicion of Down syndrome, the level of certainty of the attending physician at the time of disclosure, and the time interval between disclosure of clinical suspicion and confirmation of karyotype. Initial uncertainty and a delay in the diagnosis were detrimental to parental coping, as was premature communication of the news. Perinatal complications increased parental anxiety regarding their child's condition and future. Individual communication style of midwives and physicians was a powerful predictor of parental adaptation. Parental needs for support and information were facilitated through normalising postnatal care, ensuring privacy, and providing early access to peer support and up-to-date written information. Many parents would have appreciated access to a liaison worker.

Conclusion: The experiences of parents in this study provide practice points for improving postnatal care with minimal changes to formal service systems.

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Parents who receive an unexpected diagnosis of Down syndrome in their newborn child go through many stages of reaction and adaptation. Even if the diagnosis was suspected during pregnancy, its confirmation can be a shock. Contact with formal sources of support, information and treatment have been shown to contribute to parents' ability to cope, ultimately affecting the future wellbeing of the child and the extended family.[1-3]

Several studies have investigated sources of support received by new parents of a baby with Down syndrome, and the needs and preferences of parents following the diagnosis.[2,4-13] Some have proposed practice points for improving the delivery of care in the early stages after a baby with Down syndrome is born, to reduce the trauma associated with diagnosis.[2,7,8,11,12] Despite these recommendations, parental needs are not always met. A questionnaire-based study of the mothers of 74 children with Down syndrome born in Victoria during the period 1995–1996 showed that information and support for families was not structured and depended heavily on the commitment of the health professionals involved with the birth.[14]

We explored how formal sources of support are experienced by Victorian families with a newborn diagnosed with Down syndrome. We also documented family preferences for information and support in the early stage after diagnosis, with a view to formulating practice points for improving postnatal care.

METHODS

Eligibility criteria for families in the study were that the infant was not diagnosed with Down syndrome before birth, the infant was born between 1 January 2002 and 31 December 2004 and was living at the time of interview, and the parents were fluent in English. To reduce sampling bias, eligible participants were identified through the Victorian Birth Defects Register, rather than by a call for volunteers. For privacy reasons, invitations to participate were then sent to each mother by the physician who notified the birth to the Register. Ethics approval for the study was obtained from the Victorian Government Department of Human Services Human Research Ethics Committee.

The theoretical framework underpinning the study was a model of stress and coping developed by McConachie.[15] The model comprises two main elements that influence parental adaptation after a major stressful event — parents' individual coping styles and available resources. The role of formal sources of support (the focus of this study) is embedded in the latter.

As we wanted to explore parents' needs and experiences in detail, in-depth interviews were conducted by one of us (E E M). Interviews began with an open-ended question: Would you please tell me about the time your baby was born and when it was first suspected that all was not quite as it should be? This allowed parents to describe their experience, reflecting on issues that were important to them at the time of diagnosis. Further questions were asked by the investigator for clarification and expansion. Interviews were audiotaped and transcribed verbatim. Interpretive content analysis[16] was undertaken by systematically identifying emerging categories using NVivo 7 (QSR International, Doncaster, Vic), a qualitative research software package. Coding of categories was independently verified by one of us (V R C).

RESULTS

A total of 123 potentially eligible mothers were identified, and the physicians of 70 were traced and contacted in writing. Thirteen of these physicians informed us that the mother had moved interstate or that they did not have contact details for her. From the remaining physicians, 17 families were recruited for the study. One additional mother volunteered to participate because she had heard about the study from a third party.

Of the 18 families who participated, 4 lived in rural areas of Victoria and 14 lived in Greater Melbourne. Eight women gave birth in a private hospital and 10 did so in public hospitals. The mothers' ages ranged from 25 to 43 years, and 11 of the 18 were

younger than 35 years at the time of the birth. The children with Down syndrome were aged between 1 and 4 years at the time that interviews took place, and in nine families the child with Down syndrome was the firstborn. Seven interviews were conducted with both parents, and 11 with mothers only; the duration of the interviews was 50–70 minutes.

Influences on parental coping

Analysis of the interviews identified five major themes that related to parents' reactions at the time of diagnosis and the influence of health professionals on parents' ability to come to terms with the diagnosis.

Time interval between birth and disclosure of clinical suspicion of Down syndrome. In some instances the parents were told that the infant was suspected to have Down syndrome before the mother's immediate needs were seen to or before she held the infant for the first time. Parents felt that their baby was taken away from them before they had the chance to bond or experience the sense that they had become parents. The baby was replaced by what one mother termed a "health problem". However, parents also understood that the clinical suspicion often arose from their baby's physical appearance at birth, and they did not wish to be deceived by doctors.

Level of certainty of attending physician at time of disclosure. The disclosure of the clinical suspicion of Down syndrome was sometimes delivered with relative certainty, but on other occasions physicians waited for the cytogenetic test result (karyotype) before disclosing the confirmed diagnosis. Parents who were asked to wait for the test result were not given any information about Down syndrome while they waited, even if they requested it. Parents spoke of their increasing anxiety, expecting the worst or convincing themselves that none of this was actually happening.

Time interval between disclosure of clinical suspicion and confirmation of karyotype. If there was a low level of certainty when the suspicion of Down syndrome was first raised, the cytogenetic test result proved to be the most important factor for parents, as it determined the course of their "new lives". In some instances, parents waited 2–3 weeks for the results, whereas others received the results within 2 days. Some parents also mentioned that it was useful to receive a copy of the test report.

Communication style of health professionals. Parents' reactions were strongly influenced by how midwives and physicians communicated with them, but parents also acknowledged that the interplay between the communication style of health professionals and the family's emotional reaction was difficult to predict.

All parents responded favourably to, and remembered, health professionals who sat down by their bed, listened to them, or made a special effort to follow up on their questions. Many parents were able to quote remarks made by health professionals that marked a turning point for them.

Level of perinatal complications. Many infants in this study were delivered by caesarean section, and even infants without major comorbidities spent time in a special care nursery because of apnoeic episodes or jaundice. This affected mother–infant bonding and increased anxiety in both parents. Parents spoke of a loss of control and feeling helpless, compounded by a fear of losing their baby.

Parental support and information needs

A further five themes emerged when parents described their needs for postnatal support, including their preferences for being provided with information at an early stage.

Normalisation of postnatal care. Parents generally expected to have a relatively uneventful delivery, and they received the news that their baby had Down syndrome with great distress. Many experienced difficulties in relating to their newborn, while grieving for the child they did not have. Parents often spoke about how long it took to realise that their infant was "their baby" first and "a child with Down syndrome" second. Parents remembered health professionals who congratulated them on having a baby and treated their infant as they would any other. Some mothers also said that they experienced difficulties with breastfeeding but wanted to persist because they saw this as one of the few things they could actively do for their baby.

Privacy. Some parents expressed their need to be able to spend as much time together as possible during their hospital stay, and wished to limit their number of visitors. Most parents were offered a private room by the hospital.

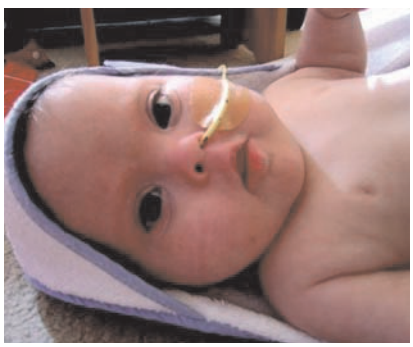
Early access to peer support. Some parents described how desperate they were to speak with another family of a child with Down syndrome, whereas others waited weeks or even months before contacting another family. Two sets of parents explained that they were hesitant to ask for access to peer support, but would have been grateful if it had been organised for them. Reasons that parents gave for speaking to another family were to hear about how life can return to normal, what the capabilities of a child with Down syndrome are, how to deal with specialist surgery, and how to help siblings cope. Some parents experienced considerable anxiety at the possibility of meeting another child with Down syndrome. In some instances, the peer-support parent brought a photo album to the visit, chronicling their child's life in a family context. Other avenues of peer support included friends, paediatricians, maternal and child health nurses, and early intervention centres. In most cases, access to peer support was facilitated through the local Down syndrome association, either via the hospital or at the parents' own initiative.

Early provision of up-to-date information. Parents' needs for information varied between individuals and according to the age of the child. Some parents bought books, and most received an information kit from the local Down syndrome association. A small number of parents relied on the Internet for information. Parents were generally discouraged by the myriad of negative information about potential comorbidities and level of developmental delay, and remembered information that emphasised the child's potential. Parents of older children explained that, in hindsight, most of the information they read in the early stages was outdated, impersonal, and did not relate to their child in particular, and that their lives took a much more positive turn than they expected. Many parents commented on how all children are different and that this is the same for children with Down syndrome — something they did not understand at first.

Access to a liaison worker. Although a liaison worker was not available to any of the parents in this study, many expressed their wish for such a person to guide them through their time in hospital and to link them with services. Parents spoke of a need for continuity of care and consistency of advice, as they explained how tired they became from telling and re-telling their story to different health professionals. Some hospitals provided social workers, but they were generally perceived as providing

Practice points for improving care for parents of a newborn with Down syndrome

- Health professionals' verbal and non-verbal communication has a potentially lifelong impact on parents.
- The effect of uncertainty of the diagnosis in the first few days after birth is considerable. Opportunities for promoting parent–infant bonding and normalising postnatal care need to be balanced with raising the clinical suspicion of Down syndrome as soon as possible.
- Confirmation of the clinical suspicion of Down syndrome should be expedited by using techniques such as fluorescent in situ hybridisation to determine karyotype.
- Maternity services should be encouraged to provide a private room for both parents during the postnatal stay.
- A designated health care worker, who can assist with coordination and navigation of the health care system, could help provide continuity of care for families with an infant with Down syndrome.
- Parents should be provided with up-to-date written information about Down syndrome and information about peer support while they are still in hospital.



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counselling that was of uncertain or limited usefulness. In cases where genetic counselling was offered, parents were reluctant to accept this as they again felt uncertain about its value. Also, the term "genetic" was often described as confusing or frightening.

DISCUSSION

This study provided insight into the experiences of parents of a newborn with Down syndrome at the time of diagnosis, and their need for support and information. Parents' tendency to remember exactly how they were first told of their child's condition and the way this framed their subsequent reactions indicate the importance of this moment. After this, parents rely heavily on their own personal resources and coping strategies, as well as formal supports.[15,17-19]

A delay in delivering this sort of news can be detrimental to parents' ability to cope, and parents wish to receive such news early.[4,5,11] However, we showed that parents are unhappy when they are told that their infant may have Down syndrome straight after birth, before the mother's immediate needs are attended to, or before they hold or see their baby, which is consistent with previous findings.[4,11] This suggests that there is a need to balance the timing of disclosure with allowing parents to welcome their newborn. Small gestures such as congratulating parents on the birth of their child were much appreciated by the parents in our study, and it is disheartening that these were rare.

Parents also expressed anxiety associated with the uncertainty of the clinical suspicion. This could be alleviated by fast-tracking karyotype confirmation using techniques such as fluorescent in situ hybridisation, and providing information about Down syndrome with sensitivity to parents' needs at the time. Although parents' reaction to the diagnosis is clearly influenced by the communication style of the paediatrician, many parents are able to differentiate between their reaction to the diagnosis and to the manner in which it was delivered.[5,20]

Training doctors in how to communicate bad news is very important and could be improved.[21,22] Current training in Victorian medical schools includes several modules spread across the course teaching students to deal with clinical scenarios that involve breaking bad news and communicating sensitively and effectively. At the postgraduate level, an apprenticeship model is vital for equipping paediatric trainees to communicate bad news. Paediatric training and continuing medical education programs have used interactive technology to discuss the best way to communicate unexpected news.[21] An example is the Communication Skills Simulation Programme at Royal Children's Hospital, Melbourne, which is open to all medical staff but targeted particularly to junior staff. It involves a workshop using videos of consultants, as well as opportunities for junior doctors to practise giving bad news with a trained actor. However, the most important training opportunities remain real-life situations, in which junior staff can observe skilled and experienced clinicians telling families honestly and compassionately that their infant has a serious disability.

Continuity of care facilitated by a liaison worker might resolve many of the other issues raised by parents in this study.[23,24] It might assist in the normalisation and continuity of postnatal care, ensure consistency of advice, and facilitate appropriate referral to a lactation consultant or peer support. However, liaison workers who support new parents of a child with Down syndrome need to feel confident about providing information in the context of a family-centred model, work with other health professionals involved in the care of the family, have up-to-date resources available, and understand the referral system for early intervention. As the birth of a baby with Down syndrome or other disability is uncommon, it might not be feasible to employ liaison workers at every hospital. A possible solution is a roving allied health professional skilled in providing the required support, such as a genetic counsellor. Although genetic counselling in Victoria is predominantly used in the prenatal setting for Down syndrome, it has been shown to be equally useful in a similar setting after a postnatal diagnosis.[25] In addition, parents' concerns regarding the term "genetic" might be alleviated by a more purposeful explanation of the genetic counsellor's role in postnatal support.

This study had some limitations. We cannot be certain that the experiences and needs of the families in our study are representative of those of most families with a newborn with Down syndrome. However, consistent themes emerged from the interviews, and it is likely that similar factors influence the postnatal experiences of other families. This study focused on parents' early needs for support and information and did not look at other predictors of parental coping and adaptation, such as individual coping strategies, family functioning, and housing, finance and employment status. Their importance should not be underestimated. Also, additional complexities exist for families from culturally and linguistically diverse backgrounds, who were not included in this study.

A positive experience with health care providers can contribute significantly to a family's emotional recovery after an unexpected diagnosis of Down syndrome in a newborn, regardless of individual coping strategies. Despite the myriad of literature on breaking bad news and communicating

with parents of a newborn with Down syndrome, a need for change continues. The experiences of parents in this study provide practice points to help health professionals improve care with minimal changes to formal service systems (Box).

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COMPETING INTERESTS

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AUTHOR DETAILS

Evelyne E Muggli, MPH, Research Officer[1]
Veronica R Collins, PhD, Senior Research Officer[1]
Catherine Marraffa, FRACP, FRCPCH, Deputy Director[2]

1 Murdoch Childrens Research Institute, Melbourne, VIC.
2 Department of Developmental Medicine, Royal Children's Hospital, Melbourne, VIC.

Correspondence: **evi.muggli@mcri.edu.au**

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