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ORIGINAL RESEARCH – QUANTITATIVE

Providing family-centred care for rare diseases in maternity services: Parent satisfaction and preferences when dysmelia is identified

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ABSTRACT

Problem and background: Dysmelia is usually detected prenatally or postnatally in maternity services. The provision of family-centred care for parents at the time of initial diagnosis is crucial to facilitate decision making, access to appropriate services, and the provision of parental care-giving, but no research has investigated parent experiences or preferences in this population.
Aims: The current research aimed to address this by investigating satisfaction with service, occurrence of signposting and preferences in this group.
Methods: Two online surveys were conducted. In the first survey ($n = 417$), parents reported whether they were offered signposting information and their level of satisfaction with the service they received when initially diagnosed. In the second survey ($n = 130$), a subgroup of participants who completed the first survey reported their preferences for signposting and health service access after diagnosis.
Findings: On average, participants were less than satisfied with the service they received and only 27% were offered signposting information. Satisfaction was higher amongst parents who had been offered signposting information. 91% of parents said they would have wanted signposting information and 67% would have wanted access to a support group.
Conclusions: There is a need to improve the family-centeredness of care when dysmelia is identified. Offering signposting information to relevant third-sector organisations may increase parent satisfaction and address parent preferences. These findings could have implications for parents of children with other rare diseases identified in maternity services.

Summary of relevance:

Problem

- Family-centred care at the time that dysmelia is identified is crucial to support parent decision making and care-giving, but no research has investigated parent experiences or preferences in this population.

What is already known

- Parents of children with other congenital differences report finding signposting information for third sector organisations beneficial.

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What this paper adds

- There is room for improving the care provided in maternity services when dysmelia is identified in babies pre or postnatally. Offering parents signposting information to relevant organisations may represent one simple step towards providing family-centred care.

1. Introduction

Rare diseases are considered to be those which affect less than 200,000 people in the United States of America (US)[1] or 1 in 2000 people in the European Union (EU).[2] Up to 8000 rare diseases exist altogether[2] and when aggregated, they affect 25 million people in the US[3] and 29 million in the EU.[2] Dysmelia refers to a group of rare diseases involving congenital limb reduction differences, occurring in around 6–8 in 10,000 births.[4–6] It is identified via ultrasound in 35–50% of cases,[5,7] and if diagnosed at this time, parents will experience uncertainty about the likely degree of disability and a potentially higher risk of foetal demise or stillbirth.[4,7] They may also need to make a decision regarding terminating the pregnancy, and termination rates have been reported to vary across specific diagnostic groups, ranging from 20% to 50%.[4–6] If identified postnatally, parents may need rapid access to information and support, in order to enable them to provide care for a child with a disability.[8]

Family-centred care can be understood as care which encourages and facilitates family support and networking, responds to the needs of families, and emotionally supports them.[9] At the time that a limb difference is identified, family-centred care is necessary to enable optimal parental decision making and care provision for infants with dysmelia. One key need for parents at this point is the identification of a specific diagnosis, as these have different associated aetiologies, symptom profiles and expected outcomes, which can influence parent decisions prenatally, and inform the healthcare needs that both they and the infant will have postnatally.[5,6] However, due to a scarcity of relevant knowledge in local health services, the identification and correct specific diagnosis of dysmelia and other rare diseases is delayed, with deleterious physical, cognitive and psychological consequences.[10] The current EU strategy to address this is to develop rare disease networks of individuals with rare diseases and experts to provide rapid access to peer-to-peer support, specialised services and high quality health care.[2] These networks constitute third-sector organisations in that they are voluntary or charitable organisations which are independent of the government, which aim to support individuals and families with rare diseases. Where the focus of individual support-groups is on providing peer-to-peer forums, specialist networks instead aim to (i) link individuals with rare diseases with both each other and also with relevant experts, (ii) liaise with and network individual support groups with each other, and (iii) develop networks internationally. There is currently no research into whether parents find these networks useful when receiving a diagnosis of a rare disease in maternity services, but research in parents of children with less rare disorders has suggested that parent networks and support groups are extremely useful, providing emotional and educational support, and helping parents to problem solve.[8,11,12] As parents of children with dysmelia need to access specialist care to identify a specific diagnosis, these networks may be particularly beneficial and support the wellbeing of infants with dysmelia, but this has yet to be established. The present research aimed to address these issues through two surveys investigating levels of parent satisfaction, prevalence of signposting (where contact details for organisations able to provide further information/support are provided), and preferences for health care provision at the time of initial diagnosis amongst parents of children with a limb difference.

There is also a need to understand factors which may influence signposting and parent satisfaction. For example, due to the greater number of people affected by the drug thalidomide during the 1960s in Europe, and the current EU strategy to support specialist networks, there is more third-sector support for dysmelia in Europe than other locations. However, it is unclear whether this has led to improved signposting and parent satisfaction in European nations than elsewhere. Similarly, research in other populations has indicated that parents prefer to know about foetal anomalies or disabilities as early as possible, as this can allow for mental preparedness for the birth,[13] but it is unclear whether early (i.e., prenatal) identification of dysmelia is associated with higher parent satisfaction than postnatal detection. The present research aimed to address these issues, and is the first that we are aware of which has investigated parental experience and preferences when rare diseases are identified in maternity services. As such, it may have wider implications for rare disease management in this setting.

The first survey aimed to assess (i) overall prevalence of signposting and level of parent satisfaction with service at the time of initial diagnosis, (ii) whether the time of initial diagnosis (prenatal v postnatal), geographical location (continent) and severity of disability (perceived severity and number of limbs affected) was associated with variation in parent satisfaction with service or occurrence of signposting, and (iii) whether being offered signposting information was associated with greater parent satisfaction with service. The second survey aimed to assess the information and services parents would have liked to be offered.

2. Methods

2.1. Design, participants and ethics

A cross-sectional survey was hosted online between July 2014 and November 2014 and advertised internationally via EDRIC (the European Dysmelia Reference Information Centre, a specialist network for individuals with dysmelia and dysmelia experts) mailing lists, the EDRIC website and social media. It was available in English, French, Italian and German languages. Parents and carers of children with dysmelia were invited to participate. A subgroup of respondents to the first survey (participants who responded to the survey between August 2014 and November 2014; $n = 130$) were invited to complete a second survey containing three additional questions addressing the services they would have liked to receive. The research was approved by the School of Psychology, University of Leeds Ethics Committee.

2.2. Surveys

The surveys were developed collaboratively with representatives from EDRIC. They were developed based on (i) knowledge gaps in the healthcare services research literature, and (ii) clinical need as identified by clinicians and parents of children with dysmelia who were associated with EDRIC.

The first survey consisted of 20 items. The current study reports finding from 9 items concerning the year and geographical location of birth, perceived severity of dysmelia (mild, moderate or severe), number of affected limbs, timing of diagnosis (antenatal v postnatal) and diagnostic category, satisfaction with experience of diagnosis (rated on a 5-point scale from 1 – Very Dissatisfied to 5 – Very Satisfied) and whether signposting occurred. The remaining 11 items

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