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Providing Family-centred Care for Rare Diseases in Maternity Services: Parent Satisfaction and Preferences when Dysmelia is Identified

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Providing Family-centred Care for Rare Diseases in Maternity Services: Parent Satisfaction and Preferences when Dysmelia is Identified

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.

Providing Family-centred Care for Rare Diseases in Maternity Services: Parent Satisfaction and Preferences when Dysmelia is Identified

Introduction

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.
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.

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 8,000 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 fetal 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 fetal 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.

Methods

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.

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 included on the survey were not relevant to the aims of the current project, and so were not included in the analysis.

The second survey contained three items asking parents whether 1) they would have liked access to sources of information (general health information, specialist health information, patients or peer networks or other), 2) they would have liked access to specific health professionals (obstetrician, paediatrician, psychologist, geneticist, plastic surgeon, specialist hand/foot surgeon, orthopaedic consultant, prostheticist, other), and 3) they would have liked to receive support from other parents who have children with limb difference or

specialised experts. For items 1 and 2 of the second survey, participants could endorse as many or few categories as applied. For item 3, participants were asked to respond 'yes' or 'no'.

Analysis Plan

Data analysis was undertaken using the Statistical Package for the Social Sciences (SPSS) version 22. As a large number of countries (n=18) and diagnostic categories (n=28) were reported, geographical location was aggregated to continents (Europe, North America and Other) and the four most common diagnostic categories (Femur Fibula Ulna complex (FFU), Poland Syndrome, Symbrachydactyly and Amniotic band syndrome) were used in the analyses.

Survey 1. Descriptive statistics were conducted for geographical location, perceived severity of disability, timing of diagnosis, specific diagnosis received, type of limb difference and the relationship of the respondent with the child with dysmelia. A series of bivariate logistic regression analyses were undertaken to identify whether signposting to organisations (outcome) varied according to time of initial diagnosis (prenatal vs. postnatal), perceived severity of disability (mild vs. moderate vs. severe), perceived satisfaction and location. Due to small numbers of cases the 'other' category in location was collapsed, and comparisons were made between Europe vs. all other locations, and North America vs. all other locations. Categorical predictors were entered as dummy variables. Ordinal regression models were conducted to investigate whether reported satisfaction with diagnostic process (outcome) varied according to perceived severity of disability (mild vs. moderate vs. severe), type of limb difference and location. Due to small numbers of cases the "other" category for limb difference was collapsed, and comparisons were made between four limbs affected vs. one affected limb and four limbs vs. other numbers of affected limbs (i.e., two or three). Also due to small numbers, the category for location was collapsed again, creating comparisons between Europe vs. all other locations, and North America vs. all other locations. A single predictor was included in each model.

Survey 2. Initially, descriptive statistics were conducted and the subgroup was compared to the remaining sample on various indicators, to test whether this sample was representative of the sample in the first survey. Due to the response key for items 1 and 2 (where as many items as applied could be checked) it was not possible to distinguish true missing data from response boxes that were left blank because they did not apply. For the purposes of this report non-response was assumed to represent the latter option, and no imputation was used.

Missing data

There were a total of $n = 452$ responses to the first survey. After excluding those who only answered two or fewer questions $n = 417$ remained. Rates of missing data ranged from 1.92% (relationship with individual) to 17.51% (diagnosis). Complete data was present for $n = 274$ individuals. Missing data was associated with several study variables (e.g., type of limb difference, language) suggesting data were Missing at Random (MAR)[14].

Missing data was handled using Multiple Imputation (MI). This approach is considered the gold-standard for managing missing data in a way that minimises bias[14]. It involved the creation of $m = 20$ datasets[15] where missing data were imputed based upon the other variables in the model (language, location, birth year, type of limb difference, diagnosis, support satisfaction, offered signposting, respondents relationship to individual, point of diagnosis, perceived severity). Values were able to vary across each imputed dataset. Analyses were then undertaken on each dataset and pooled results were calculated. This process preserved the variability in the data. A sensitivity analysis was undertaken using MI on the complete data only. No substantive differences were present. For inferential analyses (logistic and ordinal regression) only imputed data were used. Descriptive statistics are, however, provided for both imputed data and complete case data. Influential cases were screened for in the complete case data using DfBeta values and Cook's distances (values > 1 suggest an influential case[16]). Where identified, sensitivity analyses were conducted comparing the results with and without these influential cases to establish whether they made a substantial impact to the results.

Results

Descriptive Statistics. Descriptive statistics for the study variables are reported in Table 1. For Survey 1, results are provided both for the means-imputed data and the non means-imputed data. Approximately a third of participants reported receiving an initial diagnosis during prenatal screening. A majority of respondents were mothers based either in Europe or North America. Mean satisfaction with experience of diagnosis indicated that on average, participants were less than satisfied. A minority of respondents were offered signposting information to a relevant third-sector organisation. The subgroup responding to Survey 2 (n = 130) were more likely to be older, more likely to be from Europe, more likely to be fathers (as opposed to mothers) and more likely to be in the 'other' diagnostic category (compared to FFU) than the remaining sample (n = 287, Table 1).

Table 1 here

Signposting. Results of the logistic regressions are reported in Table 2. The odds of being offered signposting information for relevant organisations were greater for those who found out about the limb difference prenatally. They were greater for those in Europe compared to other continents, with those in Europe having more than twice the odds of being offered signposting information. They were also lower for those in North America compared to other continents. Perceived severity was unrelated to whether signposting was offered. Being offered signposting information was associated with greater satisfaction. No outlying influential cases were identified.

Table 2 here

Parental Satisfaction with Screening and Diagnosis. Results of the ordinal regression models are reported in Table 3. The test of parallel lines was non-significant for all models ($p > .05$) suggesting the proportional odds assumption was met in all cases. Satisfaction was significantly lower amongst those reporting four limbs affected compared to those with other types of limb difference, and there was a trend towards those with four

affected limbs reporting lower satisfaction than those with one affected limb (p = .05). Satisfaction was also lower amongst those reporting mild compared to severe perceived severity and amongst those who found out postnatally compared to prenatally.

Table 3 here

Parents Preferences

Amongst participants responding to the second survey, a desire for information on support groups and specialist treatments was particularly well endorsed (Table 4). Of the various professional groups, consultation with a psychologist was most desired within this sample. The item asking whether participants would have liked support from other parents of a child with a limb difference or to specialised experts was endorsed by a large majority of respondents.

Table 4 here

Discussion

The time of initial diagnosis is a challenging time for parents of children with a limb difference, who may need to make difficult decisions with little information if diagnosis occurs prenatally, or provide care without preparation if the diagnosis occurs postnatally. As such, the provision of family-centred care at this point is crucial to both support optimal decision making and to enable access to specialist care which could impact the long-term health of the child[10]. The present research found that on average, parents were not satisfied with their experience of receiving a diagnosis, and only 27% were offered signposting to a specialist source of support. Levels of satisfaction varied according to timing of diagnosis, with prenatal diagnosis leading to higher satisfaction, and according to level of disability, with more severe disability and higher number of affected limbs associated with lower satisfaction. Signposting occurred more than twice as often in Europe than elsewhere, and the occurrence of signposting was associated with higher satisfaction. The second survey into parent preferences suggested that a majority of parents were keen to access a specialist network (91%) and information on support groups (67%). In terms of support from

health professionals, around 40% of parents reported that they would have liked access to a psychologist and a specialist hand/foot surgeon.

The low level of reported parent satisfaction suggests that maternity services are currently failing to provide family-centred care to parents of children with a limb difference at the point of diagnosis. This finding is consistent with research into parent experience of diagnosis of fetal anomaly[17] and Down syndrome[11], where parents have also described a poor experience. This perceived shortfall in service provision was most keenly felt amongst parents of more severely disabled children, and parents whose child was not diagnosed until after birth, who reported the lowest levels of satisfaction. There has been some debate about the utility of prenatal ultrasound screening, as it is rare that treatment can occur prior to birth[18,19]. However, the present results support previous research suggesting that early identification is preferred by parents, even when no medical action can be taken[13]. The reasons for this preference were not investigated in the present study, but may be due to enabling the parents to prepare for the birth[20]. No evidence was found to suggest a higher level of parent satisfaction in Europe than elsewhere, which was contrary to expectations. Due to the greater historical population level of limb difference caused by thalidomide, and the concomitant higher level of third sector support (i.e., the specialist network EDRIC), service provision could have been expected to be best here. There are several possible explanations for this finding, including the possibility that third-sector organisations such as EDRIC have not been effective. However, given that signposting was found to occur more frequently in European nations it seems that third-sector organisations have indeed been effective in raising awareness and offering parental support. Instead, these results may indicate a need for further integration between health services and third-sector organisations, in order for maternity services make changes in service provision in response to service-user preferences.

These results suggest that being offered signposting to a specialist network can help to provide a better parent experience. Not only was signposting associated with higher satisfaction when it did occur, but 91% of parents said they would have wanted to be offered

signposting information. This is consistent with previous research indicating that after news of a fetal anomaly, parents are keen to gain more information quickly and will utilise sources other than their health service, including the internet[17,20]. In line with our predictions, signposting was found to be higher in Europe than elsewhere. The findings also suggested that signposting was more likely to occur when the diagnosis was made prenatally, highlighting a particular problem with signposting in postnatal services. In the EU, where specialist networks are currently receiving funding, these results demonstrate a need for maternity services to provide information to staff about specialist networks and to encourage signposting. In the US and elsewhere, these results demonstrate this same need, but also advocate for greater investment in such networks.

The current research provided clear results regarding parent preferences. In general, a higher number of parents wanted access to information and non-health professional support (support groups) than to specific health professionals. In particular, a majority of parents said that they wanted access to a specialist network (91%) and support groups (67%). In terms of support from health professionals, around 40% of parents reported wanting access to psychology services and to a hand/foot surgeon. Around a third of parents wanted access to a geneticist, orthopaedic consultant or prostheticist. These findings are consistent with previous research suggesting that peer-to-peer support is important to a majority of parents of children with a disability[11,12], but that parents' desire to access professional support is more varied. Previous research has indicated that some parents are keen to access as much information from professionals as possible, whereas others prefer to process the news of an anomaly more gradually[20]. Together with the present results, these findings suggest that parents should be offered access to a wide range of services, but the uptake of these will depend upon individual parent's coping styles and preferences.

Limitations

These findings must be considered in the light of two main limitations. First, the group of participants included in this study were self-selecting, responding to advertisements via the European Dysmelia Reference and Information Centre (EDRIC) and social media.

Because of this, they are more likely to be parents who have engaged with sources of support. This may have led to larger numbers of parents endorsing a desire for a network or support group than a study of consecutive parents identified via a hospital register might have. However, the current study represented over 400 parents of children with dysmelia. Given the rarity of dysmelia (6-8 per 10000), gaining such a large sample size via a systematic study of births would require over 570,000 births to be followed. Furthermore, previous research in groups where a pregnancy abnormality was found has found no difference in the data from parents who were recruited via a hospital and those recruited via a network[21].

Second, the research was not conducted immediately following the birth of the children, which may have influenced responses due to distortion in memories over time. However, mean year of birth was 2006, and most parents responding to the survey were describing experiences within the past decade. Furthermore, allowing for this passage of time i) was more ethical, as parents were less likely to still be experiencing distress related to the event, and ii) may mean that responses were less influenced by diagnosis-related distress, and may have benefited from time for reflection.

Conclusions

The present research is the first to investigate parent satisfaction, signposting and preferences regarding the prenatal or postnatal diagnosis of dysmelia, or indeed any rare disease in maternity services. Providing family-centred care at this time is important for supporting parental decision making, and to enable parents to provide care for infants with dysmelia. The results suggested low levels of satisfaction and signposting (27%), but that signposting was higher in Europe where there is greater third-sector support for dysmelia. Prenatal diagnosis, being offered signposting information, and lower levels of disability were associated with higher levels of satisfaction. A majority of parents wanted to be offered signposting information to a specialist dysmelia network (91%) or a parent support group (67%). Around 40% of parents also reported a desire to access psychology services or to see a specialist hand-and-foot surgeon. This research is the first to investigate parent

experiences of diagnosis of a rare disease in maternity services, and provides evidence that further studies using large birth cohorts is warranted.

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Table 1

Descriptive Statistics for Survey 1 and comparison of Subgroup who Responded to Second Survey to those Responding Only to the First Survey

Variable		Imputed data[a] n (%)	Completer data[b] n (%)	Participants responding to both surveys n (%)	Participants only responding to Survey 1 n (%)	Group difference OR
Continent	Europe	223 (53.3)	209 (55.3)	119 (91.5)	103 (35.9)	1
	North America	176 (42.1)	160 (42.3)	9 (6.9)	167 (58.2)	0.05**
	Other	19 (4.5)	9 (0.02)	2 (1.5)	17 (5.9)	0.11*
Severity	Mild	117 (28.1)	101(27.7)	31 (24.6)	86 (30.0)	1
	Moderate	211 (50.6)	189 (51.8)	68 (52.3)	144 (50.2)	1.31
	Severe	89 (21.3)	75 (20.5)	32 (24.6)	57 (19.9)	1.54
Timing of Diagnosis	Postnatally	256 (61.4)	245 (60.9)	84 (64.6)	172 (59.9)	1
	Prenatally	161 (38.6)	157 (39.1)	46 (35.4)	116 (40.4)	0.80
Diagnosis	FFU	95 (22.8)	69 (20.1)	10 (7.7)	84 (29.3)	1
	Poland syndrome	42 (10.1)	32 (9.3)	14 (10.8)	28 (9.8)	5.04
	Symbrachydactyly	85 (20.4)	69 (20.1)	8 (6.2)	77 (26.8)	0.89
	Amniotic band syndrome	80 (19.2)	68 (19.8)	16 (12.3)	63 (22.0)	2.69
	Other	117 (28.1)	106 (30.8)	82 (63.1)	34 (11.8)	25.06**
Type of limb difference	Four limbs affected	42 (10.1)	40 (10.2)	8 (6.2)	34 (11.8)	1
	Two limbs affected	266 (63.8)	255 (65.1)	23 (17.7)	243 (84.7)	0.38*
	One limb affected	93 (22.3)	91(23.2)	92 (70.8)	1 (0.03)	[c]
	Other	16 (3.8)	6 (1.5)	7 (5.4)	9 (3.1)	4.20
Relationship with affected person	Mother	364 (87.1)	359 (87.8)	103 (79.2)	261 (90.9)	1
	Father	34 (8.1)	33 (8.1)	22 (17.0)	12 (4.2)	4.55**
	Other	20 (4.8)	17 (4.2)	6 (4.6)	14 (4.8)	1.08
Signposted	Yes	111 (26.6)	103 (24.7)	49 (37.7)	62 (21.6)	2.17**
		Mean	Mean (SD)	Mean	Mean	OR
Birth year		2006.23	2006.67 (8.33)	2005.13	2006.73	0.98
Satisfaction		2.59	2.61 (1.35)	2.49	2.64	0.92

** p < .05; ** p < .001*

[a] These are the descriptive statistics once adjustments had been made for missing data. These data were used to conduct all inferential statistics.

[b] These are the descriptive statistics for the participants who completed this items on the questionnaire, before adjustments for missing data were made

[c] Comparison could not be made as frequencies too low

Note: MI can produce pooled frequencies that are not whole numbers. These have been rounded to whole numbers in the current table. MI only produces pooled means not SD.

Table 2

Bivariate Logistic Regressions Predicting whether Participants were Offered Signposting Information (significant factors presented in bold)

		95% Confidence interval	
Predictor	OR	Lower limit	Upper limit
Satisfaction with service[b]	**1.67****	**1.40**	**1.99**
Location (Europe)			
Other[a]	1	-	-
Europe	**2.40****	**1.40**	**4.12**
Location (North America)			
Other[a]	1	-	-
North America	**0.40****	**0.24**	**0.67**
Time of diagnosis			
Postnatal[a]	1	**-**	**-**
Prenatal	**1.69***	**1.07**	**2.66**
Perceived severity			
Mild[a]	1	-	-
Moderate	1.02	0.59	1.74
Severe	0.74	0.37	1.50

* $p \leq .05$; **$p \leq .01$; OR = Odds Ratio; [a] Reference category; [b] Five-point scale (and as such, no reference category was included).

Table 3

Ordinal Regressions Predicting Perceived Satisfaction (significant factors presented in bold)

		95% Confidence interval	
Predictor	OR	Lower limit	Upper limit
Location (Europe)			
Other[a]	1	-	-
Europe	0.84	0.56	1.27
Location (North America)			
Other[a]	1	-	-
North America	1.09	0.75	1.60
Time of diagnosis			
Prenatal[a]	1	-	-
Postnatal	**0.48****	**0.33**	**0.69**
Perceived severity			
Mild[a]	1	-	-
Moderate	0.93	0.61	1.41
Severe	**0.41****	**0.23**	**0.72**
Type of limb differences			
Four limbs affected[a]	1	-	-
One limb affected	**2.33***	**0.99**	**5.47**
Two or three affected limbs	**3.14****	**1.40**	**7.04**

* $p \leq .05$; ** $p \leq .01$; OR = Odds Ratio; [a] reference category

Table 4

Information and Services Parents Would Have Liked to Receive

	n (%)
Information on raising child with limb difference	60 (46.15)
Information on specialist treatment	66 (50.77)
Expert directory	57 (43.85)
Information on support groups	87 (66.92)
To consult with Obstetrician	6 (4.62)
To consult with Paediatrician	26 (20.00)
To consult with Psychologist	53 (40.77)
To consult with Geneticist	35 (26.92)
To consult with Plastic Surgeon	10 (7.69)
To consult with Specialist Hand/Foot Surgeon	51 (39.23)
To consult with Orthopaedic Consultant	38 (29.23)
To consult with Prostheticist	38 (29.23)
Support from other parents or specialised experts	110 (90.90)