JIDR Journal of Intellectual Disability Research
Published on behalf of mencap and in association with IASSID

Journal of Intellectual Disability Research doi: 10.1111/jir.12151

VOLUME 59 PART 5 pp 474–486 MAY 2015

A tale worth telling: the impact of the diagnosis experience on disclosure of genetic disorders

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Abstract

Background Research suggests children with genetic disorders exhibit greater coping skills when they are aware of their condition and its heritability. While the experiences parents have at diagnosis may influence their decision to disclose the diagnosis to their children, there is little research into this communication. The aim of the current study was to examine the relationship between the diagnosis experience and the disclosure experience for parents of children with developmental disorders of a known genetic aetiology: parents of children with 22q11.2 deletion syndrome (22q11DS) were compared with a group of parents with children affected with other genetic diagnoses, with a similar age of diagnosis (e.g. fragile X syndrome) and a group where diagnosis generally occurs early (i.e. Down syndrome).

Method The sample comprised 559 parents and caregivers of children with genetic developmental disorders, and an online survey was utilised. Items from the questionnaire were combined to create variables for diagnosis experience, parental disclosure experience, child's disclosure experience, and parental coping and self-efficacy.

Results Across all groups parents reported that the diagnosis experience was negative and often accompanied by a lack of support and appropriate information. Sixty-eight per cent of those in the 22q11DS and 58.3% in the Similar Conditions groups had disclosed the diagnosis to their child, whereas only 32.7% of the Down syndrome group had. Eighty-six per cent of the Down syndrome group felt they had sufficient information to talk to their child compared with 44.1% of the Similar Conditions group and 32.6% of the 22q11DS group. Parents reported disclosing the diagnosis to their child because they did not want to create secrets; and that they considered the child's age when disclosing. In the 22q11DS and Similar

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Conditions groups, a poor diagnosis experience was significantly associated with negative parental disclosure experiences. In the Similar Conditions group, a poor diagnosis experience was also significantly associated with a more negative child disclosure experience.

Conclusions As expected this study highlights how difficult most parents find the diagnosis experience. Importantly, the data indicate that the personal experiences the parents have can have a long-term impact on how well they cope with telling their child about the diagnosis. It is important for clinicians to consider the long-term ramifications of the diagnosis experience and give the parents opportunities; through, for instance, psychoeducation to prepare for telling their child about the diagnosis. Further research is warranted to explore what type of information would be useful for parents to receive.

Keywords 22q11.2 deletion syndrome and families, diagnostic disclosure in genetic conditions, disclosure of diagnostic information to children, parental perceptions of diagnostic experience, velo-cardiofacial syndrome and families

Introduction

It can be a shocking and stressful experience for parents to receive the news that their child has a multisystem genetic disorder (Hallberg *et al.* 2010; Metcalfe *et al.* 2011), and it can be similarly difficult for parents to decide if and what to tell the child about the diagnosis. Research suggests that children with genetic disorders exhibit greater coping skills when they are aware of their condition and its heritability (Hughes *et al.* 2002; Tercyak *et al.* 2002; McConkie-Rosell *et al.* 2009; Metcalfe *et al.* 2011). Despite this positive finding, many parents choose not to disclose to their children or to only partially disclose (Gallo *et al.* 2005; Metcalfe *et al.* 2008). Although the communication experiences around the disorder potentially have a large impact on family functioning (Rolland 1994), there is little research into the communication of genetic conditions, both from professionals to parents ('diagnosis') and from parent to child ('disclosure'). The absence of advice regarding disclosure is particularly problematic when intellectual functioning is affected, as different approaches may be needed for parents to effectively communicate the diagnostic information to their children (Faux *et al.* 2012).

One such genetic disorder is 22q11.2 deletion syndrome (22q11DS; also known as velo-cardio-facial syndrome), which occurs in approximately 1:4000 live births making it one of the most common microdeletion syndromes (Wilson *et al.* 1994). The syndrome is associated with characteristic facial features, congenital heart defects and abnormalities of the palate (McDonald-McGinn *et al.* 1999). The behavioural phenotype is characterised by executive dysfunction (Bish *et al.* 2005), attention deficits (Niklasson *et al.* 2005), social impairments (Shashi *et al.* 2012), autism spectrum disorder features (Fine *et al.* 2005) and anxiety disorders (Fung *et al.* 2010) and there is a significantly increased risk of mood (Green *et al.* 2009) and psychotic disorders compared with the general population (Murphy *et al.* 1999). Other features of the disorder can include increased risk of infection (Jawad *et al.* 2001), neonatal hypocalcaemia (Kitsiou-Tzeli *et al.* 2005) and recurrent otitis media (Dyce *et al.* 2002). People with 22q11DS often have a borderline intellectual functioning or mild to moderate intellectual disabilities; however, the majority have the intellectual capacity to understand the implications of their genetic condition (Faux *et al.* 2012). Despite 22q11DS being one of the more common genetic developmental disorders, the diagnosis is often delayed until later childhood or adulthood as the level of awareness of the syndrome is low among professionals. 22q11DS can be difficult to recognise because of large inter- and intra-familial symptomatic variability (Shprintzen 2008), with some people having few or even none of the more well-known features (such as cardiac or palatal anomalies) of the syndrome but nonetheless having the deletion

Anecdotally, many parents of children with 22q11DS report a traumatic experience around the time of diagnosis (e.g. Hallberg *et al.* 2010) and this may in turn have an impact on if, how and when parents choose to divulge the diagnosis to the child (Forrest *et al.* 2003). A diagnosis can provide relief and comfort for parents in terms of understanding the origin of symptoms, which can facilitate treatment and prognosis, despite the sorrow and grief about their child's 22q11DS (Hallberg *et al.* 2010;

Costain *et al.* 2011). Even though putting a name and/or origin to the symptoms can be a relief, the experience of receiving a diagnosis is frequently distressing. Parents of children with various conditions (including 22q11DS) have identified the need for adequate and understandable information from health professionals both at the time of diagnosis and in the years following the diagnosis (Green & Murton 1996; Baird *et al.* 2000; Hallberg *et al.* 2010). Qualitative research findings on genetic disorders and cancer in children suggest that when parents have a more negative diagnosis experience, the communication around the syndrome and/or the decision to inform the child is negatively influenced; however, this is yet to be quantitatively supported (Young *et al.* 2003; Hallberg *et al.* 2010).

People typically choose to disclose a genetic disorder to their children because they feel an obligation to do so, the child has shown interest in their condition, to explain medical interventions or to ensure the child does not feel shame (Hughes *et al.* 2002; Gallo *et al.* 2005; Metcalfe *et al.* 2008; Faux *et al.* 2012). From research pertaining to various genetic conditions, it appears that full disclosure is typically associated with more adaptive parental coping skills including an open communication style and a focus on problem-solving (Tercyak *et al.* 2001; Metcalfe *et al.* 2011). Non-disclosure can occur in 22q11DS because parents feel unclear on how and what to tell their child (Faux *et al.* 2012). Hence, parents have identified the need for professional advice regarding developmentally appropriate methods of disclosing (Metcalfe *et al.* 2011; Faux *et al.* 2012). Although advice from professionals may help parents cope emotionally (Metcalfe *et al.* 2008), Gallo *et al.* (2005) reported that 80% of parents to children with various disorders received no professional advice regarding the method of disclosure.

The purpose of the current study was to investigate the diagnosis and disclosure experiences in families where a child has 22q11DS. In order to elucidate these experiences, parents of children with 22q11DS were compared with parents of children with other genetic developmental disorders with similarly complex phenotypes and characteristics ('Similar Conditions'), such as impaired cognition, life-long symptoms and delayed diagnoses (i.e. tuberous sclerosis, Williams, Prader–Willi and fragile X syndromes), as well as a group of parents of children with Down syndrome where the diagnosis tends to happen earlier. In addition, Down syndrome is distinguished from these other syndromes because of its relatively high incidence, high public awareness and well-recognised characteristic facial features.

It was predicted parents who disclosed would have had a more positive diagnosis experience compared with those who did not disclose. It was also hypothesised that the diagnosis and disclosure experiences would be more positive for the Down syndrome group, as health professionals are well educated in this condition and can provide more information. The nature of the relationship between the parental diagnosis experience and disclosure experience for both parents and children was examined for each of these groups. It was expected that a positive diagnosis experience would result in a more positive disclosure experience for both the parent and the child. Finally, it was predicted that self-efficacy and coping skills would mediate the relationship between the diagnosis experience and the disclosure experience.

Methods

Participants

The sample comprised 559 parents and caregivers (subsequently referred to as 'caregiver respondents') of children with genetic developmental disorders (22q11DS $N = 193$, Down syndrome $N = 122$, fragile X syndrome $N = 34$, Williams syndrome $N = 48$, tuberous sclerosis $N = 111$ and Prader–Willi syndrome $N = 51$). Participants were required to be 18 years or older, have an adequate English reading level and be a parent or caregiver to at least one child with one of the aforementioned conditions.

Measures

Because of a lack of published questionnaires investigating diagnosis and disclosure experiences, the authors created a survey based on a review of the literature, qualitative interviews with family members and clinical experience. The survey also contained items from questionnaires such as the 'STIGMA Shout Survey' (Corry 2008), 'Inventory

of parent's experiences' (Crnic *et al.* 1981), the 'Coping Inventory for Stressful Situations – Adult' (Endler & Parker 1990) and the 'Being A Parent – Mother' questionnaires (Johnston & Mash 1989). The questionnaire contained a mixture of restricted questions (multiple choice), quantitative rating scales and open-ended qualitative questions. The survey was piloted with a sample from the target population in Australia and Israel prior to full-scale administration to ensure that the questions were relevant, appropriately worded and that the questionnaire was not too long. Subsequently, the survey was modified to accommodate the changes recommended (e.g. removing and/or rewording questions).

The resulting survey contained 109 items. For the purpose of the current study items from the questionnaire were combined in order to create grouping variables for *diagnosis experience* (e.g. 'How would you rate the amount and quality of information from the health professional about the syndrome at the time of diagnosis'; 1 = Satisfactory, 7 = Unsatisfactory), *parental disclosure experience* (e.g. 'How prepared did you feel to have the conversation about the diagnosis with your child?'; 1 = Unprepared, 7 = Well prepared), *child's disclosure experience* (e.g. 'Did your child show feelings of distress as a result of talking about the diagnosis?'; 1 = Very distressed, 7 = No distress) and *parental coping and self-efficacy* (e.g. 'I meet my own personal expectations for expertise in caring for my child'; 1 = Strongly disagree, 7 = Strongly agree) for each caregiver respondent. Therefore, higher scores for the parental disclosure experience, child's disclosure experience, and parental coping and self-efficacy indicate a more positive experience. However, for the diagnosis experience, higher scores mean a more negative experience. An additional variable was created for *disclosure decision*; that is, 'have you told your child about the diagnosis?' with response options of yes or no.

In order to test the psychometric properties of these composite variables, internal consistency was examined through item-to-total correlations [minimum criterion = 0.5 (Hair *et al.* 1998)] and inter-item correlations [minimum criterion = 0.3 (Hair *et al.* 1998)]. Then, principal components analysis confirmed if the items constituted one underlying construct [unidimensionality assumed when only one component had an eigenvalue >1, with all loadings > 0.5 (Hair *et al.* 1998)]. The constructs were examined for reliability using Coefficient Cronbach's alpha [acceptable $\alpha = 0.60$ (Hair *et al.* 1998)]. All composite variables met these criteria with the exception of *child's disclosure experience*, where reliability was poor ($\alpha = 0.484$).

Procedure

To overcome small sample sizes and to maximise response rates a web-based approach was utilised. A website was created that contained information on the objectives of the research and hyperlinks to the questionnaire [hosted through the online survey software, *Zoomerang* (http://www.zoomerang.com)], where potential participants could read the information statement and begin the questionnaire. The study website link and a brief blurb were posted on Facebook pages, newsletters, blogs and websites for neurodevelopmental disorders. Of the 99 groups and pages contacted, 39 agreed to advertise the study, 2 actively declined and 58 did not respond. The blurb and link were posted at regular intervals to ensure they remained visible and accessible. In addition, 40 (7.16%) of the participants were recruited through clinics in Clinical Genetics, in order to get a sample more representative of the population. The survey was conducted with the understanding and consent of the participants, and received ethical approval from the University of Newcastle's Human Research Ethics Committee.

Data analysis

Differences between caregiver respondents who disclosed and those who did not disclose were compared through a multivariate analysis of variance (MANOVA), with factors identified throughout the literature as affecting the choice (i.e. diagnosis experience, parental coping and self-efficacy, condition type and child's age) as dependent variables and disclosure decision as the independent variable. To examine whether the diagnosis experience and disclosure experiences (for parent and child) differed between 22q11DS, Similar Conditions and Down syndrome, a series of one-way analysis of variances (ANOVAs) were conducted with condition

type as the independent variable and each of diagnosis experience, parental disclosure experience, child disclosure experience, and parental coping and self-efficacy as dependent variables. The relationship between the parental diagnosis experience and disclosure experiences was tested through correlations for each condition. A mediation model was proposed to examine the hypothesis that self-efficacy and coping skills would mediate the relationship between the diagnosis experience and the disclosure experience.

Relevant statistical assumptions were tested and found to be acceptable. However, significant levels of skewness were identified across several variables such as *parental coping and self-efficacy* for both 22q11DS and Similar Conditions groups. Because of the large sample size (i.e. 559 participants), parametric tests were utilised. However to ensure accuracy, confirmatory non-parametric tests were used to ensure the reliability of the statistical analysis. An alpha value of 0.05 was used in all analyses. There was missing data from the variables diagnosis experience, parental disclosure experience, child disclosure experience, and parental coping and self-efficacy (6.62%, 11.63%, 10.2% and 15.74% respectively). A listwise deletion approach was taken, as it appeared to be missing at random. However, caution must be taken when interpreting the results because data may be missing for reasons the researchers are unaware of.

Results

Participant demographics (see Table 1)

The vast majority of caregiver respondents across all groups were female (22q11DS: 91%, Down syndrome: 93.1%, Similar Conditions: 90.7%) and were married or in a *de facto* relationship (22q11DS: 83.2%, Down syndrome: 85.4%, Similar Conditions: 85.6%). Around half of the respondents were living in North America, had completed at least an

Table 1 Participant demographics

Participant demographics	22q11DS *N*	Down syndrome *N*	Similar Conditions *N*
Female	171 (91%)	108 (93.1%)	225 (90.7%)
Total *N*	188	116	248
Married/*de facto*	158 (83.2%)	99 (85.4%)	214 (85.6%)
Total *N*	190	116	250
Living in North America	82 (42.7%)	68 (58.6%)	154 (62.1%)
Total *N*	192	116	248
Completed at least an undergraduate university degree	87 (46%)	58 (50%)	127 (51.6%)
Total *N*	189	116	246
Reported an average income	81 (44.3%)	48 (42.9%)	103 (43.6%)
Total *N*	183	112	236
Diagnosis experience*			
With partner	77 (41.4%)	55 (49.1%)	89 (36.8%)
Alone	50 (26.9%)	20 (17.9%)	51 (21.1%)
Total *N*	186	112	242
Child's age at diagnosis			
Prenatal	3 (1.6%)	21 (18.8%)	12 (5%)
Birth–6 months	68 (36.8%)	90 (80.4%)	90 (36.8%)
7–< 24 months	22 (11.9%)	1 (0.9%)	59 (24.4%)
2–5 years	43 (24.3%)	0 (0%)	66 (27.3%)
6–> 18 years	46 (24.9%)	0 (0%)	15 (6.2%)
Total *N*	185	112	242

* Remainder of caregiver respondents were with family members, relatives, friends and/or the child.
Similar Conditions includes tuberous sclerosis, Williams syndrome, fragile X syndrome and Prader–Willi syndrome.
22q11DS, 22q11.2 deletion syndrome.

undergraduate university degree, and reported an average income. Commonly, children with Down syndrome were diagnosed between birth and 6 months (80.4%) or prenatally (18.8%). Many of those with 22q11DS and Similar Conditions were also diagnosed at birth to 6 months of age (36.8% and 36.8% respectively); however, a fair proportion were not diagnosed until 2–5 years (24.3% and 27.3% respectively). Over a quarter of the caregiver respondents in the 22q11DS group were alone at the diagnosis, and 41.4% were with a partner. The remaining respondents were with the child; or the child plus family members, friends or relatives. However, in the Down syndrome group, almost 50% of participants were with a partner at diagnosis and only 17.9% were alone. The remainder in this group were with the child and/or family members, friends or relatives. Medians for the diagnosis experience of 4–6 were recorded for 22q11DS and Down syndrome groups, and 5–7 for the Similar Conditions group on the relevant, individual Likert scale items (1 = positive, 7 = negative).

Contingency table analyses were conducted to establish whether there was a significant relationship between condition type and each of gender, marital status, education level and income. No significant relationships were found. Chi-squared analyses demonstrated a significant relationship between (a) age at diagnosis and condition type, χ^2 ($DF = 14$, $n = 539$) = 183.58, $P < 0.001$, with the Down syndrome group over-represented in the prenatal to 6-month period and under-represented in the 7–< 24 months 2–5 years and 6–10 years age groups. The 22q11DS group was under-represented in the prenatal and over-represented in 6 to 10 year age groups. The Similar Conditions group was under-represented in the birth to 6 months and over-represented in the 7–< 24 months and 2–5 years age groups; (b) disclosure decision and condition type, χ^2 ($DF = 2$, $n = 510$) = 34.32, $P < 0.001$, with the Down syndrome group over-represented in non-disclosure and under-represented in the disclosure group; this was reversed in the 22q11DS group; (c) country of residence and condition type, χ^2 ($DF = 8$, $n = 556$) = 45.37, $P < 0.001$, with the 22q11DS group over-represented in Israel and under-represented in North America; and the Down syndrome group under-represented in Israel; and (d) age of the child at disclosure and condition type, χ^2 ($DF = 8$, $n = 245$) = 28.05, $P < 0.001$, with the 22q11DS group over-represented in the 16 years or older group and the Down syndrome group over-represented in feeling that the child has always known of their diagnosis. That is, although they had told of or discussed the child's Down syndrome with them, they felt there was no specific age of disclosure.

The majority of caregiver respondents in the 22q11DS and Similar Conditions groups had disclosed the diagnosis to their child (68% and 58.3% respectively). However, 67.3% of the Down syndrome group had not disclosed. Commonly, caregiver respondents disclosed because they did not want to create secrets and did not disclose when they felt their child was too young. Largely, caregiver respondents in the 22q11DS and Similar Conditions groups disclosed when the child was 5–10 years of age, yet many parents in the Down syndrome group felt there was no specific disclosure age, the child had always known (40%). Eighty-six per cent of the Down syndrome group felt they had sufficient information to talk to their child compared with only 44.1% of the Similar Conditions group and 32.6% of the 22q11DS group. Over 50% of caregiver respondents in the Similar Conditions group had not been given advice about disclosure, nor had 35% of those in the 22q11DS group and 13.3% in the Down syndrome group. The child's disclosure experience, as rated by the caregiver respondents, was generally positive for all condition types, with Likert scale medians for the individual questions ranging from 5 to 6 for 22q11DS and Similar Conditions groups, and 6 to 7 for the Down syndrome group (1 = negative experience, 7 = positive experience). Parental coping and self-efficacy questions had medians ranging from 4 to 6 in the 22q11DS groups, 5 to 7 in the Down syndrome group and 4 to 7 in the Similar Conditions group on the relevant Likert scales; where 1 = poor coping and self-efficacy, 7 = good coping and self-efficacy.

Experimental outcomes

To examine potential factors affecting the disclosure decision across all conditions, a between subjects MANOVA was conducted with disclosure decision

(yes or no) as the independent variable and four dependent variables [i.e. diagnosis experience, parental coping and self-efficacy, condition type, and child's age (divided into age groups where 1 = 0–2 years, 2 = 3–5 years, 3 = 6–12 years, 4 = 13–17 years, 5 = 18+ years)], see Table 2. With the use of Pillai's criterion, the combined dependent variables were significantly related to disclosure decision, $F_{4,421} = 53.88$, $P < 0.001$. Univariate analysis found that the mean child's age group for participants who had disclosed ($M = 3.66$, SD = 1.09) was significantly higher than for those who had not disclosed ($M = 2.06$, SD = 1.18). No other significant results were identified.

Then, a series of one-way ANOVAs were performed to investigate differences between the three condition type groups in terms of diagnosis experience, disclosure experiences (parental and child's), and parental coping and self-efficacy, see Table 2. They revealed that there was no significant effect for condition type in terms of diagnosis experience ($F_{2,519} = 1.68$, $P = 0.187$). However, the Down syndrome group ($M = 24.13$, SD = 3.40) displayed significantly more positive parental disclosure experiences than both 22q11DS ($M = 19.49$, SD = 4.75) and Similar Conditions ($M = 20.75$, SD = 5.14) groups ($F_{2,219} = 8.85$, $P < 0.001$). Caregiver respondents in the Down syndrome group ($M = 11.88$, SD = 1.90) also had more positive child disclosure experiences than both 22q11DS ($M = 10.39$, SD = 2.30) and Similar Conditions ($M = 10.43$, SD = 5.14) groups ($F_{2,224} = 4.34$, $P = 0.014$). However, because of the unequal group sizes (i.e. fewer caregiver respondents in the Down syndrome group had disclosed compared with those in the 22q11DS and Similar Conditions groups),

Table 2 MANOVA and ANOVA results for disclosure decision and (a) diagnosis experience, (b) parental coping and self-efficacy, (c) condition type and (d) child's age

MANOVA	Disclosure decision	Mean	Std. deviation	*N*	*F*	Sig.
Diagnosis experience	Yes	19.16_a	5.90	237	0.26	0.872
	No	19.25_a	6.02	189		
Parental coping and self-efficacy	Yes	39.11_a	5.40	237	1.179	0.278
	No	38.46_a	7.04	189		
Condition type	Yes	2.10_a	0.94	237	1.997	0.158
	No	2.22_a	0.78	189		
Child's age group	Yes	3.66_a	1.10	237	210.392	<0.001
	No	2.06_b	1.18	189		
ANOVA	**Condition type**	**Mean**	**Std. deviation**	***N***	***F***	**Sig.**
Diagnosis experience	$22q11DS_a$	19.58	5.67	178	1.680	0.187
	Down $syndrome_a$	18.27	6.05	107		
	Similar $Conditions_a$	19.18	5.91	237		
Parental disclosure experience	$22q11DS_a$	19.49	4.75	91	8.845	<0.001
	Down $syndrome_b$	24.13	3.40	24		
	Similar $Conditions_a$	20.75	5.14	107		
Child's disclosure experience	$22q11DS_a$	10.39	2.30	93	4.337	0.014
	Down $syndrome_b$	11.88	1.90	26		
	Similar $Conditions_a$	10.43	2.58	111		
Parental coping and self-efficacy	$22q11DS_{a,b}$	38.63	5.49	166	4.173	0.016
	Down $syndrome_b$	40.41	6.20	97		
	Similar $Conditions_a$	38.32	6.39	208		

Means in the same cell that do not share the same subscripts are significantly different, $P < 0.01$.
22q11DS, 22q11.2 deletion syndrome.

the results must be interpreted with caution. Finally, the Down syndrome group ($M = 40.41$, SD = 6.20) had higher mean coping and self-efficacy than caregiver respondents in the Similar Conditions ($M = 38.31$, SD = 6.39) group ($F_{2,468} = 4.17$, $P = 0.016$).

Finally, to examine the nature of the relationships (if any) between the diagnosis experience and disclosure experiences for parent and child, correlations were carried out. A small negative correlation was found between diagnosis experience and parental disclosure experience for the 22q11DS group, $r = -0.221$, $P = 0.038$ and the Similar Conditions group, $r = -0.313$, $P = 0.001$; see Table 3. That is, (because of the scaling of the survey items) caregiver respondents who had a more negative experience around the diagnosis also had a negative experience at disclosure; however, this relationship

Table 3 Correlations for diagnosis experience, parental and child disclosure, and coping and self-efficacy

	Diagnosis experience	Parental disclosure experience	Child disclosure experience	Parental coping and self-efficacy
22q11DS diagnosis experience	1			
	.	–	–	–
	178			
22q11DS parental disclosure experience	–0.221	1		
	0.038	.	–	–
	88	91		
22q11DS child's disclosure experience	0.028	0.369	1	
	0.790	<0.001	.	–
	90	86	93	
22q11DS parental coping and self-efficacy	–0.091	0.353	0.193	1
	0.252	0.001	0.067	.
	159	87	91	166
Similar Conditions	1			
Diagnosis experience	.	–	–	–
	237			
Similar Conditions	–0.313	1		
Parental disclosure experience	0.001	.	–	–
	106	107		
Similar Conditions	–0.216	0.529	1	
Child's disclosure experience	0.023	<0.001	.	–
	110	101	111	
Similar Conditions	–0.063	0.100	0.093	1
Parental coping and self-efficacy	0.368	0.328	0.355	.
	206	97	101	208
Down syndrome	1			
Diagnosis experience	.	–	–	–
	107			
Down syndrome	0.164	1		
Parental disclosure experience	0.444	.	–	–
	24	24		
Down syndrome	0.058	0.410	1	–0.082
Child's disclosure experience	0.780	0.052	.	0.722
	26	23	26	21
Down syndrome	–0.185	–0.082	0.248	1
Parental coping and self-efficacy	0.073	0.722	0.254	
	95	21	23	97

22q11DS, 22q11.2 deletion syndrome.

only accounted for about 7% to 8% of the variance. No significant correlation was found between the diagnosis experience and the child's disclosure experience in the 22q11DS group $r = 0.028$, $P = 0.79$. However, a small negative correlation was found between the diagnosis experience of the caregiver respondent and the child's disclosure experience for the Similar Conditions group $r = -0.216$, $P = 0.023$; again, accounting for only about 5% of the variance. There was no significant correlation between diagnosis experience of 22q11DS or Similar Conditions and the potential mediator of parental coping and self-efficacy ($r = -0.091$, $P = 0.252$ and $r = -0.063$, $P = 0.368$ respectively) hence the mediation model was not conducted. No significant correlation was found between diagnosis experience and parental disclosure experience or child's disclosure experience for the Down syndrome group ($r = 0.16$, $P = 0.444$; $r = 0.06$, $P = 0.78$; see Table 3).

Discussion

We investigated the parental experience of having a child diagnosed with a genetic developmental disability and if the parental experience affected their decision to disclose the diagnosis to the child. Across conditions, most caregiver respondents rated the experience of being told the diagnosis as negative. That is, based on the questions in the diagnosis experience scale; they found it stressful, felt extremely worried, felt they initially had a poor understanding of the syndrome, and the amount and quality of information they received from health professionals was unsatisfactory. Caregiver respondents in the Down syndrome group disclosed to their child earlier and felt more prepared to do so than 22q11DS and Similar Conditions groups. However, close to 70% of caregiver respondents in the Down syndrome group had not disclosed. This high rate of parents not disclosing to their children may be due to type of questions asked in the questionnaire; with many parents proposing that although they did not specifically tell their child, they had gradually learnt about the diagnosis over the years. Alternatively, the timely diagnosis may allow for the caregiver respondents who disclosed to adjust and reflect while still disclosing relatively early in the child's life. Further research with a greater proportion of disclosing caregiver respondents is required.

Contrary to expectations, across all caregiver respondents the diagnosis experience did not differ between those who did and did not disclose, nor was there any significant differences in coping and self-efficacy skills or condition type between those who did and did not disclose. A potential explanation for this effect is that regardless of the type or time of diagnosis, the caregiver respondents found the situation equally traumatic because of the challenges their child would face. This is not to say the diagnosis should be avoided. The importance of a diagnosis must be recognised, as parents have also reported positive outcomes, such as relief, as a result of the news (e.g. Hallberg *et al.* 2010; Costain *et al.* 2011). Rather, the focus should be on reducing the impact of potentially distressing news by meeting caregivers' needs. The diagnosis experience is often perceived as more positive if the knowledge is provided in a calm, supportive manner and with a partner or close friend present (Green & Murton 1996; Baird *et al.* 2000). Further, when a diagnosis is provided genetic counsellors can facilitate the process of gathering a support network, by referring caregivers to relevant local and online support groups; of which there are many for each condition type investigated in this study. In the case of a 22q11DS diagnosis, health services can provide and explain the international clinical practice guidelines for 22q11DS to parents (Bassett *et al.* 2011).

As expected, the Down syndrome group had more positive parental and child disclosure experiences compared with both 22q11DS and Similar Conditions; as Down syndrome is well known compared with the other disorders included in the study. Interestingly, the majority of the caregiver respondents of children with Down syndrome had not disclosed to their child (whereas the opposite was true in both 22q11DS and Similar Conditions groups). The current age of children who had been disclosed to was significantly higher than those who had not been told. This is somewhat expected as caregiver respondents of 22q11DS children have identified concerns about the language to use when disclosing (Faux *et al.* 2012), which may abate as the child grows. Research on various genetic conditions has demonstrated that parents believe non-disclosure is emotionally

protective; because of fears of causing anxiety, inferiority and changed self-perception in their child (Hughes *et al.* 2002; Tercyak *et al.* 2002; Claes *et al.* 2003; Forrest *et al.* 2003; Gallo *et al.* 2005; Metcalfe *et al.* 2008; McConkie-Rosell *et al.* 2009). Despite these concerns, there is little empirical evidence that emotional damage eventuates as a result of disclosure. Rather, children who know about their genetic diagnosis often show improved emotional resilience (Metcalfe *et al.* 2011). Although evidence is lacking for children with 22q11DS, in families with open communication, children with genetic disorders exhibit increased coping skills, improved attitude to their condition, fewer psychosocial issues and reduced stress levels (e.g. Metcalfe *et al.* 2011; Plumridge *et al.* 2011). Indeed, qualitative accounts have shown that non-disclosure creates secrecy that negatively impacts family cohesion, causing children to feel stressed, frustrated, resentful and anxious (Claflin & Barbarin 1991; Metcalfe *et al.* 2008; Plumridge *et al.* 2011). A positive disclosure experience for children occurs when children are informed about their diagnosis in a timely manner, taking cognitive development into account, with the opportunity for continued discussion (Metcalfe *et al.* 2011; Plumridge *et al.* 2011; Faux *et al.* 2012).

Unexpectedly, there was no relationship between the diagnosis experience and parental or child disclosure for the Down syndrome group, perhaps because of the comparatively small number of disclosing caregiver respondents. Participants' strong coping and self-efficacy skills may have also prevented the diagnosis experience from affecting disclosure. However, as predicted, there was a relationship between the diagnosis experience and parental disclosure for each of 22q11DS and Similar Conditions groups. There was also a relationship between the diagnosis experience and the child's disclosure experience for the Similar Conditions group only. Contrary to predictions, these relationships were not mediated by parental coping. Although qualitatively identified in the choice to disclose, diagnosis experience may not be a particularly significant factor in the disclosure decision for the wider population. Also, the literature used to guide the predictions was based on conditions with different prognoses and implications. Further, there may be factors with an influence on disclosure decision that were not examined in the study, such as familial relationships, thus there is a need for ongoing scientific examination.

The current study had a number of limitations; in particular the study's web-based approach did not allow an investigation of the sample's representativeness of the wider population. An ascertainment bias may restrict the findings as the participants were primarily sourced through online support groups. Members of support groups often actively seek information and may have received advice from others on disclosure. The biased sampling was also demonstrated through the high levels of self-efficacy and coping exhibited by the caregiver respondents. Despite these limitations, the web-based approach was chosen to allow international accessibility, flexibility for participants, and to improve on the small sample sizes found in previous research. A small proportion of participants ($N = 40$) were also recruited through genetic clinics in order to reduce the impact of these sampling issues. The sample was predominantly female and thus the results may be biased towards a female perspective. Another limitation is that the child's response to disclosure was based on their parent's perception of the situation. However, similar research has shown continuity between parent and child responses (Costain *et al.* 2011). Also, it is possible the study attracted people who had strong feelings about the subject, perhaps because of their own diagnosis or disclosure experiences. This issue may have contributed to the lack of findings for the planned mediation analyses. Further, a small proportion of caregiver respondents from the 22q11DS and Similar Conditions groups may have been affected by the disorder themselves. This could impact their expectations of a diagnosis for their child, how they reacted to the diagnosis, and whether they chose to disclose. To some extent it may explain the large proportion of disclosing parents in both these groups, as compared with the Down syndrome group. An exclusive examination of diagnosis and disclosure experiences when the parent has been diagnosed with the same condition is an avenue for future research. The use of retrospective self-report data also may have affected

participants' answers; however, research has shown that parents tend to remember these experiences accurately (e.g. Carr 1988). A final limitation that should be considered is our measurement tool. We endeavoured to explore the scale's initial psychometric properties; yet some of the findings may have been influenced by unknown measurement error or rater biases. While we feel confident in our findings, we are sensitive to the issue that the rating scale may have influenced our findings.

Conclusions

This is the first known study to quantitatively investigate the relationship between diagnosis and disclosure experiences. As the child's age was the only significant examined factor in the disclosure decision, it would be advantageous to determine other variables that may impact the decision to disclose, such as life events, sibling relations, child's temperament and other key family variables (e.g. socioeconomic status, family size). Also, separate analysis for each Similar Condition could increase understanding of the effect condition type has on diagnosis and disclosure experiences.

From a clinical perspective, the current research has implications for medical professionals and genetic counsellors, such as meeting parents' informational and emotional needs at diagnosis, as well as throughout changes in the child's developmental stages. Further, if future research identifies variables that influence the disclosure decision, such as parental coping and self-efficacy, it may provide a foundation for parental training. It is important to examine the factors affecting the disclosure decision as communication patterns are related to the family's resilience (e.g. McConkie-Rosell *et al.* 2009). The diagnosis experience remains a distressing event for many parents and caregivers and thus is worthy of inclusion in future research. Genetic testing and receiving a diagnosis should not be avoided; rather, health professionals need to be aware of the impact their diagnosis can have on the parent's and thus the family's emotional well-being.

Acknowledgements

The authors would like to thank all the people and organisations that chose to volunteer as participants in the study and shared their experiences so freely. Without you we cannot do this type of research.

Conflict of interest

The authors declare that there is no conflict of interests regarding the publication of this article.

Source of funding

Funding for the current study was provided by the NIMH International Consortium on Brain and Behavior in 22q11.2 Deletion Syndrome (grant number 5U01MH101722-02) and the Binational Science Foundation (grant number 2011378).

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Accepted 25 June 2014