Toward a conceptual and methodological shift in craniofacial research

Abstract

Objective: To date, research investigating the psychosocial impact of craniofacial conditions has produced conflicting findings. The aims of this article were to critically evaluate the challenges limiting progress in the field, and to offer alternative perspectives and approaches to guide future research and practice.

Design: A comprehensive evaluation of papers exploring adjustment to congenital craniofacial conditions was conducted. Methodological approaches and underlying conceptual issues were identified and summarised.

Results: The conceptual limitations identified include: inherent challenges pertaining to the multifactorial and fluctuating nature of adjustment; a lack of consensus regarding the primary constituents of a positive outcome; scant use of appropriate models and theories; and a predominant focus on ‘deficits’ over ‘strengths’. The methodological shortcomings identified include: bias within samples; biomedical inclusion/exclusion criteria; inconsistency in measurement; a relative absence of the patient perspective; variability in data analysis and interpretation; and the failure to capitalise on knowledge from a broader range of health fields and disciplines. Findings are believed to be relevant to all disciplines involved in craniofacial research and practice.

Conclusions: The literature remains fraught with difficulties similar to those identified 25 years ago. The present article proposes that steps toward a conceptual and methodological shift are needed in order to construct a comprehensive understanding of adjustment to craniofacial conditions, and to address the key unanswered questions important to all stakeholders.

Key words: cleft; craniofacial; research; measurement; outcome; method
**Introduction**

Congenital craniofacial anomalies (CFAs), such as cleft lip and/or palate (CL/P), Treacher Collins Syndrome, Crouzon Syndrome, Apert Syndrome and craniosynostosis affect thousands of new-born infants each year worldwide. Although surgeries to repair the primary malformations take place during early infancy, patients are normally expected to engage in a multidisciplinary treatment pathway throughout childhood and into adulthood. Despite vast improvements in surgical techniques and service provision for those born with CFAs in recent decades, medical interventions can rarely ‘remove’ or ‘cure’ the anomaly entirely.

In parallel, CFAs can pose a number of psychological challenges for those affected and their families. Having an unusual appearance, as well as possible hearing and speech complications can invite staring, questions and comments from others (Rumsey & Harcourt, 2004). These noticeable characteristics may make it more difficult for a child with a CFA to integrate with their peers (Hearst, 2007), particularly during adolescence, when the emphasis on appearance, romantic relationships and a sense of social ‘belonging’ becomes heightened (Liossi, 2003). In the longer-term, research has suggested that individuals affected by CFAs may be less successful than peers in relation to education and employment, wait longer to get married or to form a long-term relationship, be less likely to have their own children and be at risk of poor mental health (Ramstad et al., 1995a; 1995b; Marcusson et al., 2001; Danino et al., 2005; Yttri et al., 2011).

The potential for CFAs to impact upon many domains of life, and the importance of facilitating psychological adjustment to these conditions is now more widely acknowledged. Paradoxically, psychological support to facilitate coping and resilience among affected individuals and their families still trails behind other aspects of care (Mouradian, 2001). To date, research investigating the impact of CFAs has failed to capture a comprehensive picture of psychological adjustment. While several studies report individuals with CFAs to experience more difficulties than their unaffected peers, others report those with CFAs to demonstrate adjustment scores which are equal to, or better than norms or comparison groups (e.g. Turner et al., 1998; Hunt et al., 2005). To further complicate this picture, many positive consequences of the condition are now often reported (Eiserman, 2001; Baker et al., 2009; Berger & Dalton, 2009; Kramer et al., 2009; Feragen et al., 2010; Stock et al., 2016). As well as indicating a high degree of individual variation, such conflicts within the literature are likely due to methodological disparity.
In 1991, a review of the current state of the field prompted Strauss and Broder to discuss the move away from a biomedical model toward a broader, health-based, interdisciplinary social science model. Within this review, a variety of methodological challenges were highlighted, and the authors concluded that a new direction in craniofacial research was necessary. Twenty-five years later, two reviews pertaining to psychological adjustment to CL/P (Stock & Feragen, 2016) and to other congenital CFAs (manuscript in preparation) have drawn attention to a number of difficulties which continue to limit progress within the field. Regrettably, it seems as though many of the challenges described by Strauss and Broder (1991) are still relevant today.

These observations bring the ongoing challenges of craniofacial research into sharp focus. In order to move forward, it is necessary to re-evaluate the current state of the research and to consider potential future directions. The aims of the present article are therefore to comprehensively summarise the key conceptual and methodological challenges prevalent within the literature, and to offer alternative perspectives and approaches with the intention of providing guidance for future research and clinical practice.

**Conceptual issues**

*The nature of adjustment*

Historically, the focus of treatment for appearance-altering conditions, much like many other health conditions, was to repair the physical anomaly. Today, adjustment to an appearance-altering condition is understood to be multifaceted, involving a complex interplay of physical, cultural, psychological and social factors (Turner et al., 1998; Endriga & Kapp-Simon, 1999; Nelson et al., 2012; Rumsey & Stock, 2013; Norman et al., 2015). In response to the challenges associated with appearance-altering conditions and their treatment, some individuals experience a range of transitory difficulties, while others report problems which persist over time (Turner et al., 1998; Beaune et al., 2004; Rumsey & Stock, 2013). Further, adjustment is liable to fluctuate over time and in accordance with different life events, experiences and contexts (Warschausky et al., 2002; Nelson et al., 2012; Rumsey & Stock, 2013). When taken together, psychological adjustment appears to be a concept which is inherently difficult to capture in a comprehensive way.

*The constituents of a positive outcome*
Another conceptual limitation stems from an overall lack of consensus regarding what should be seen as the major constituents of a positive outcome for the CFA population. A range of overlapping and competing concepts are referred to, many of which are poorly defined and used inconsistently across studies (Endriga & Kapp-Simon, 1999; Eiserman, 2001; Rumsey & Stock, 2013; de Queiroz Herkrath et al., 2015). A number of reviews have also drawn attention to the lack of knowledge regarding longer-term outcomes for those affected by CFAs (Mouradian, 2001; Yazdy et al., 2007; Wehby & Cassell, 2010; Rumsey & Stock, 2013; Roberts, 2014). Without a clear consensus regarding the desired ‘end point’ for patients and of the factors which may contribute to these outcomes, research is likely to remain disparate.

A lack of models and theories

Another challenge identified in the literature refers to the lack of appropriate models and theories (Endriga & Kapp-Simon, 1999; Eiserman, 2001; Rumsey & Stock, 2013; Liddle et al., 2015). Although some models and/or theories have been posed (e.g. Baker et al., 2009; Berger & Dalton, 2009), such suggestions are rarely extended or replicated using different data (Broder, 1997; Endriga & Kapp-Simon, 1999; Eiserman, 2001; Hunt et al., 2005; Baker et al., 2009; Liddle et al., 2015), restricting the ability to test theories and models in the context of CFAs. A thorough understanding of psychological adjustment is likely to draw upon differences in aspects of several theoretical approaches; however, until a consensus in approach has been reached, coherence will be difficult to achieve.

Deficits and strengths

In recent years, research has begun to move away from a disease-prevention oriented approach toward one which is increasingly directed toward maximising quality of life (Eiserman, 2001; Mouradian, 2001). Although efforts to capture this feature of adjustment are visible in recent research (Feragen, 2012), aspects of resilience in relation to CFAs are still poorly understood and are not often measured or discussed within the literature (Eiserman, 2001; Strauss, 2001; Beaune et al., 2004; Hunt et al., 2005; Nelson et al., 2012; Rumsey & Stock, 2013). In addition, many of the challenges faced by individuals with CFAs throughout life are equally applicable to the general population (Turner et al., 1998; Eiserman, 2001; Rumsey & Stock, 2013; Holmbeck & Aspinall, 2015), yet authors often ‘apologise’ when significant pathological differences between patients with
CFAs and control groups are not identified (Eiserman, 2001). Rather than ‘pinning’ any emotional distress on the condition (Rumsey & Stock, 2013), or indeed searching only for positive aspects, a balance between the two must be reached.

Methodological challenges

Sampling

A methodological challenge often highlighted within CFA research is the lack of large and representative samples (e.g. Endriga & Kapp-Simon, 1999; Hunt et al., 2005; Collett & Speltz, 2007; Wehby & Cassell, 2010; Richman et al., 2012; Rumsey & Stock, 2013). This difficulty is exacerbated if the population includes subgroups which are considered ‘harder to reach’, such as those belonging to an ethnic minority group, those reporting lower socioeconomic status and/or patients who no longer regularly engage with CFA services (including a large proportion of the adult population). Without access to large datasets, subgroups are likely to be neglected or misrepresented (Broder, 1997; Collett & Speltz, 2007; Zeytinoglu & Davey, 2012; Rumsey & Stock, 2013; Feragen et al., 2014). Equally, potentially confounding variables, including diagnosis, age/developmental level and gender become extremely difficult to investigate effectively (Turner et al., 1998; Collett & Speltz, 2007; Richman et al., 2012; Antonarakis et al., 2013; de Queiroz Herkrath et al., 2015). This is especially true when diagnoses are extremely rare and/or participation rates are low, resulting in samples which consist of a diverse mix of craniofacial conditions (da Costa et al., 2012; Plomp et al., 2016; manuscript in preparation). In addition, explanations of sample acquisition and sample characteristics can often be vague, and researchers are largely reliant on those patients who are most willingly to participate (Broder, 1997; Knight et al., 2014).

Inclusion/exclusion criteria

The selection of inclusion/exclusion criteria has typically been driven by biomedical parameters, and as a result, those with severe developmental and/or neurological difficulties are excluded from many CFA studies (Warschausky et al., 2002; Wyszynski et al., 2006; Rumsey & Stock, 2013; Feragen et al., 2014). While the driver for this is that restrictive selection criteria ensures homogeneity within samples, the results can become
skewed if this process is not implemented in a uniform way (Wyszinski et al., 2006; Feragen et al., 2014; Knight et al., 2014). In addition, few authors specify their exclusion criteria in sufficient detail for the implications of their sampling strategy to be assessed (Feragen et al., 2014; Maliepaard et al., 2014). A further unfortunate consequence of this targeted approach to research is that little understanding of those who are excluded is gained, despite the likelihood that these individuals are the most vulnerable (Feragen & Stock, 2014).

**Inconsistency in measurement**

Due in part to the lack of consensus regarding constructs and outcomes, a plethora of different measures have been applied within the field of CFAs. Almost all review articles have commented on the quantity of measures available, coupled with a lack of consistency and continuity across studies (e.g. Hunt et al., 2005; Klassen et al., 2012; Antonarakis et al., 2013; Maliepaard et al., 2014). Being a relatively young field, researchers often modify existing scales, and/or create new measures in order to study their construct of interest (Maliepaard et al., 2014). No standardised condition-specific measures are currently available (Broder, 2001; Klassen et al., 2012) and the development of a new scale is an extremely lengthy and demanding process (Krawczyk et al., 2012). Confusion also exists as to which measures are sufficiently validated, since studies often do not provide enough information regarding the properties of the measures they have used (Klassen et al., 2012). Together, these measurement issues render comparisons between studies extremely difficult. The number and/or length of the measures used within a given study is also a concern, whereby the complexity of adjustment needs to be accounted for without over-burdening participants (Rumsey & Stock, 2013; Stock, Hammond et al., in press).

**The patient perspective**

Many of the processes involved in adjustment to an appearance-altering condition are largely subjective and thus studies should seek to illuminate the issues and experiences which are of importance to patients and their families (Nelson, 2009). Currently, the patient perspective is not always captured in CFA studies, some of which remain dominated by reports and interpretations collected from third parties (Hunt et al., 2005; Nelson, 2009; Klassen, 2012). Qualitative research has the potential to offer powerful insights into the experience of living with an appearance-altering condition, yet there is a relative paucity of qualitative work being conducted
(Eiserman, 2001; Mouradian, 2001; Nelson, 2009; Nelson et al., 2012; Rumsey et al., 2013; Sharif et al., 2013; Liddle et al., 2015; manuscript in preparation). This is surprising in studies on rare craniofacial conditions, where larger samples are, as per definition, difficult to reach, and thus qualitative investigations would be ideal. The absence of qualitative work is especially noticeable when compared to other fields of health research (Nelson, 2009) and is particularly required in the absence of agreed quantitative measures (Hunt et al., 2005).

Analysis and interpretation

Another issue pertinent to progressing the research field is the analysis and interpretation of the research data that is collected. Of primary concern is the lack of reference to appropriate control and comparison groups (Turner et al., 1998; Hunt et al., 2005; Collett & Speltz, 2007; Wehby & Cassell, 2010; Antonarakis et al., 2013; Knight et al., 2014; Liddle et al., 2015). In addition, it can be difficult to ascertain whether comparison groups are representative of the general population and whether controls are consistently ‘matched’ (Broder, 1997; Hunt et al., 2005). Research findings are often primarily descriptive and presented as ‘statistically significant’ without also discussing corresponding effect sizes and the resulting applied clinical implications (Collett & Speltz, 2007; de Queiroz Herkrath et al., 2015). Further, Eiserman (2001) cautions against the simple comparison of affected versus non-affected samples, since groups of patients with a particular condition are unlikely to be homogenous. Studies may report patients with CFAs to have ‘poorer’ scores than those exhibited by the control or comparison group, yet those scores may still be within the normal range (Berger & Dalton, 2009). In some cases, mean scores may be chosen to replace missing data, creating a statistical problem when dealing with small sample sizes (see Geels et al., 2008).

Integrated working

A final observation is that many researchers in the field of CFAs tend to work within the silos of their respective disciplines, and an understanding of the interactions between the various affected domains of life is therefore limited (Turner et al., 1998; Nelson, 2009; Rumsey & Stock, 2013; Knight et al., 2014). Further, it is rare for professionals from other closely related fields, such as sociology, social policy, nursing and health services to be consulted (Nelson, 2009). Much more attention has been given to the area of CL/P, relative to other CFAs (Roberts & Mathias, 2012), and much of the research relating to the latter is outdated (Roberts &
Mathias, 2012; Fischer et al., 2014). Similarly, little research has broadened understanding by examining the potential overlap between CFAs and other chronic health conditions (Nelson, 2009; Rumsey & Stock, 2013).

**Discussion**

The aims of the present paper were to provide a critical synthesis of the key conceptual and methodological challenges prevalent within the literature and to offer alternative perspectives and approaches, with the intention of providing guidance for future research and clinical practice. Findings are believed to be applicable to all disciplines involved in craniofacial care.

In order to address the challenges raised and to advance research in this field, a number of suggestions are made. First, overarching conceptual frameworks are needed to guide research in terms of key constructs and interposing factors to be explored and developed (see ARC, 2009; Klassen et al., 2012; McCarthy et al., 2012; Stock, Hammond et al., in press). Frameworks should be broad, and include both generic and condition-specific constructs, in order to distinguish between normative levels of concern and the intricacies of the condition itself. Such an approach could provide a better understanding of how the cleft and its treatment has the potential to interact with, exacerbate or allay processes relating to life stages and experiences (see Lansdown et al., 1997). Equally, a framework approach should encompass elements of resilience and positive growth, as well as emotional distress and problems of adjustment, in order to capture a balanced perspective of adjustment and to support our understanding of the continuum of distress and resilience. Concurrently, research methodologies and measures which complement and inform the frameworks must be implemented consistently. Scales would ideally possess adequate psychometric properties, along with clear applications to clinical practice. For the utility of measures, models and theories to be tested and refined, replication and/or expansion of studies using the same measures is necessary, and authors should be transparent about the methodological procedures applied. A clear priority for the field over the next decade is for teams around the world to achieve consensus on the measures which best capture all of the elements of the consensus framework. The additional challenge of applicability to other countries and cultures and language translation should not be underestimated.
Second, and in order to do justice to the fluctuating nature of adjustment, there is a pressing need for long-term research across the entire lifespan. Prospective, longitudinal studies should be implemented to identify when and how particular issues become pertinent and to highlight implications for age-appropriate care (Turner et al., 1998; Broder, 2001; Mouradian, 2001; Webby & Cassell, 2010; Nelson et al., 2012; Richman et al., 2012; Rumsey & Stock, 2013; Sharif et al., 2013; Knight et al., 2014; Holmbeck & Aspinall, 2015). Although longitudinal research is notoriously difficult and time-consuming to establish and maintain (Rumsey & Harcourt, 2005, pp.41-42), the benefits of undertaking such work are likely to outweigh the challenges, so long as these challenges are openly acknowledged and discussed. Additionally, longitudinal research can be undertaken on both a large (Feragen & Stock, 2016; Stock, Humphries et al., in press 2015) and small (Murray et al., 2008; Hentges et al., 2011; Smith et al., 2014) scale. In cases where longitudinal work is not feasible, studies should utilise clearly defined age groups to avoid developmental stages and treatment phases becoming confounding variables.

A third key issue is the need for an increase in the level of patient involvement in CFA research. Collecting the perspectives of multiple informants in order to triangulate data from different sources is encouraged, although the patient’s view should always be included. The number of qualitative studies in the field remains relatively low (Stock & Feragen, 2016; manuscript in preparation), possibly because qualitative approaches are not well understood and/or lesser value is placed on them (Nelson, 2009). Where little is known about a subject, or if exploring a topic from a new perspective, qualitative data can provide insight and form a basis for future quantitative work. Additionally, when quantitative studies produce conflicting findings, qualitative approaches may help to interpret these discrepancies. One step further would be to combine quantitative and qualitative approaches in the form of mixed methods studies; an approach which could be well suited to the multidimensionality and complexity of adjustment to CFAs (Dures, 2012). Additionally, novel approaches, such as online, arts-based and visual methods hold promise and offer an exciting and engaging alternative to methods which are better established (Harcourt, 2012). In considering how to further promote the patient voice, an increasing emphasis on patient involvement in both research and practice is becoming apparent. Patient and Public Involvement (PPI, also referred to as User Involvement) requires the engagement of participants in activities at every stage of the research process, from determining research priorities through to
the dissemination of findings (INVOLVE, [www.invo.org](http://www.invo.org)), and is now often a prerequisite for research funding applications. Although PPI in the field of CFAs is still in its infancy, the amount that can be learned through patient involvement is considerable, and thus a move toward long-term PPI strategies within CFA research is regarded as essential (Bates, 2012).

Fourth, an increased effort to obtain large and representative samples would reduce the potential for misleading or inconclusive results, and could be achieved through national and international multicentre collaboration (Broder, 1997; Turner et al., 1998; Endriga & Kapp-Simon, 1999; Collett & Speltz, 2007). On a broader scale, an environment in which the delivery of health care and applied research are more fully integrated could yield many benefits for both sides. Further, the utilisation of existing data originally derived from clinical audit and/or case records has the potential to be used for research purposes (see Ruiter et al., 2009; Burnell et al., 2014; Feragen et al., 2015). Larger samples, along with a more inclusive approach to recruitment, would allow for the investigation of individual variation, potentially interacting variables and clinically important and vulnerable subgroups.

Finally, the way in which data is interpreted is crucial. Studies should seek to include a control or comparison group where possible, and be clear about the group’s characteristics and how control participants were recruited. Furthermore, and in order to distinguish between condition-specific issues and those which also apply to the general population, relevant normative data, as well as cut-off scores where available, should be referred to irrespective of whether a control or comparison group has been included. In the wider research field, further discussions are taking place as to whether statistical significance alone is enough to draw meaningful conclusions (e.g. Sullivan & Feinn, 2012). Arguably, the clinical significance is equally, if not more relevant, and effect sizes should always be included in the reporting of statistical results. Additionally, the researcher’s chosen approach and skill level has a large impact on the data that are collected, and on how that data are analysed, interpreted and presented in the context of other literature (Rumsey & Harcourt, 2005, pp. 37-38). Reflexivity is a valuable means of creating greater transparency and quality within research (Finlay & Gough, 2008). Additionally, there is a need to draw upon broader models, theories and information from related fields and disciplines to accelerate our acquisition of knowledge.
Summary and conclusions

This article has identified and summarised a wide range of conceptual and methodological challenges limiting current progress in craniofacial research and practice. Many of the points raised in current article are far from novel; yet the literature appears fraught with issues similar to those identified 25 years ago (Strauss & Broder, 1991). Particularly when compared to other areas of health research, the field of CFAs is trailing behind. The present articles proposes that steps toward a conceptual and methodological shift are needed in order to gather a comprehensive understanding of adjustment to CFAs, and to address the key unanswered questions important to all stakeholders (see Petit-Zeman & Cowan, 2013). The current article has offered suggestions for the future of the field, specifically: an appreciation of the wider context and broader experiences of the individual; consensus and consistency in relation to key constructs and measures of relevance; the implementation of longitudinal studies and/or the use of clearly defined age groups; an increase in patient involvement and improved integration of the patient perspective; the collection of large and representative samples which allow for analysis of subgroups and interposing variables; a balanced perspective of adjustment which encompasses strengths as well as deficits; and a need to draw upon knowledge acquired within other disciplines and areas of health research.

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