Long-term and multidisciplinary treatment pathways: Experiences among patients and families affected by a congenital craniofacial anomaly

Background

“I’ve had countless different appearance-altering operations in my life, and no one treating me has ever asked me how I feel about it or helped me to prepare myself for a change in appearance.”
Adult born with a craniofacial anomaly (Norwegian pilot study)

“It’s not just surgery and fixing... it’s everything that goes with it”
Mother of two sons, both born with a craniofacial anomaly (UK)

A congenital craniofacial anomaly (CFA) is a broad term used to describe a wide range of diagnoses, such as craniosynostoses, Treacher Collins, or Crouzon syndrome, to list a few. Prevalence rates and characteristics are known to vary widely across the different CFAs (Orphanet Report Series), affecting the function and the appearance of the head and face. Depending on the condition and its associated malformations, early surgical intervention, as well as ongoing multidisciplinary treatment to ‘correct’ the anomaly is often required throughout childhood and into adulthood.

In addition to the intricacies and complexities of treatment, a CFA has the potential to impact upon several domains of life (Feragen, 2012). Following a diagnosis, parents must process a wealth of new information, come to terms with their baby’s unusual appearance, cope with various health complications, and allow their new-born to undergo reconstructive surgery (Nelson et al., 2012). Although treatment can improve appearance over time, the ‘visible difference’ will not be removed entirely. As the child grows up, their ‘visible difference’ may invite unwanted staring, comments, questions and teasing (Rumsey & Harcourt, 2004), which can in turn affect their ability to form meaningful social relationships, and impact upon their emotional wellbeing (Feragen & Stock, 2016; 2017; Hunt et al., 2005; Stock & Feragen, 2016). Irrespective of the cause, extent or severity of the visible difference, looking “different” can have a profound impact in a society with a massive emphasis on appearance and “looks” (Rumsey & Harcourt, 2007). If not addressed, social anxiety and dissatisfaction with appearance can lead to anxiety and depression, in addition to detrimental health behaviours (Kanayama et al., 2006; Stice & Shaw, 2003). Appearance concerns are also known to impact on health care decision–making and could influence choices regarding aesthetic and reconstructive surgery in adolescence and adulthood, a social and health economic burden (Stice, 2002; Rumsey & Harcourt, 2004; Williamson & Rumsey, 2017). Surgical treatment pathways are well-established for patients with a CFA (routine follow-up and specialised treatment teams), yet much less is known about psychological impacts and patients’ psychological support needs.

Young people with congenital visible conditions are expected to engage in extensive and ongoing multidisciplinary treatment pathways, including surgical and medical interventions aiming at diminishing a difference that may be visible to others. Despite the long-term and complex nature of a demanding treatment pathway, little research has explored the treatment experiences of individuals with CFAs from the patients’ own perspective. A recent literature review of psychological adjustment to CFAs (Feragen & Stock, 2017), revealed that in the last 20 years, only 11 articles have focused on treatment experiences as their area of interest. Some of these studies explored patients’ satisfaction with treatment outcomes after surgery.
yet have not explored how patients experience the process of treatment. Hence, little is known about treatment and decision making experiences, emotional processes, or the consequences of appearance-altering surgery on the formation of personal identity, or perceptions of the self from the patients’ perspective. Clinical experience indicate that multidisciplinary consultations may be overwhelming for the patients and lack autonomy in relation to treatment decisions. Further, most existing studies are quantitative in nature, and based upon the views of parents and/or clinicians alone (Feragen & Stock, 2017). Given the well-known discrepancies between parent, clinician and patient views of an ‘optimal’ outcome (Kunz et al., 2014) and the importance of including patients’ subjective perspectives in research (Nelson 2009), these are important limitations of the research to date. Among the 41 studies included in the most recent review of the CFA literature (Feragen and Stock, 2017), only four Norwegian studies were identified. None of these Norwegian studies explored aspects of treatment from the perspectives of patients, parents, or that of the health professionals involved in their care.

Where previous research has sought the views, the treatment journey has been described by some as ‘physically and psychologically demanding’ (Stavropoulos et al., 2011). Two studies, one on adults with Treacher Collin syndrome (Beaune et al., 2004), and the other on adults with Crouzon syndrome (Stavropoulos et al., 2011), pointed to patients’ need for information about the diagnosis, preparation for future treatment and surgery, and for health professionals to be communicative, to listen, and to treat patients like people and not as a medical condition. Adults with Crouzon (Stavropoulos et al., 2011) discussed their desire for appearance-altering treatment, particularly during the adolescent years, yet also described the extreme burden involved in such treatment (Stavropoulos et al., 2011). Although treatment can result in enhanced wellbeing, changes in appearance may also trigger crises of identity if not managed appropriately by the clinical team (Stavropoulos et al., 2011; Tevik and Feragen, 2015). This “psychological ambivalence” towards surgical treatment has rarely been investigated, in spite of its potential impact on the development of self-perceptions and identity.

Patients across a range of other surgical procedures have reported dissatisfaction and regret with their decision to undergo surgery because it did not meet their expectations (Hamilton et al., 2013; Saban and Penckofer, 2007), whereas little is known on this subject about patients with CFAs. Several interventions aiming at enhancing communication between health professionals and patients about the consequences of treatment have been developed (Stacey et al., 2012; 2017; Stevenson et al., 2004). One of those, a simple paper-based intervention, the PEGASUS (Patient’s Expectations and Goals: Assisting Shared Understanding of Surgery; Harcourt et al., 2015) was developed by the Centre for Appearance Research (Bristol, UK), to help adult patients and health professionals elicit expectations of surgery, and set patient-centered goals. The intervention is easy to administer and implement, and encourages the patient to conceptualise their expectations of surgery, and to express these in the consultation with the aim of setting shared goals and promoting concordance between patient and surgeon, so that they approach treatment as a shared endeavor (Harcourt et al., 2015; Stevenson et al., 2004). Taking an active role in preparing for discussions with the medical health professionals empowers patients and reduces the risk of them being overwhelmed by unexpected issues and emotions during the consultation. An active disclosure and discussion of patients’ expectations is also important for the surgeon, enabling him or her to decide the extent to which expectations are realistic and to take steps to address unrealistic anticipations if necessary. Shared decision making, the process of involving patients in the treatment process, is now enshrined in Norwegian policy documents.
Ideally, treatment decisions should combine the physician’s medical expertise and the patient’s personal priority expertise (Mulley et al., 2012). Focused interventions that encourage patients to prepare for, and actively engage in consultations, are effective at improving satisfaction with health outcomes (Health Foundation, 2012; Thompson and Sunol, 1995). One study demonstrated that patients felt more confident in physicians communicating in a more patient-centered manner, which increased the likelihood of following the physician’s recommendation (Saha and Beach, 2011). Research has also shown that even experienced and specialised health professionals may be unaware of patients’ expectations and priorities, and may therefore find an intervention tool such as the PEGASUS helpful in improving communication and shared decision making (Harcourt et al., 2015).

In Norway, centralised and multidisciplinary care has been implemented for the treatment of CFAs. The Norwegian National Unit for Craniofacial Surgery is responsible for the treatment of all patients affected by rare CFA in the country. As a result, research can be conducted on large and representative samples. Most existing research on CFAs, has been carried out among children and adolescents with cleft lip and palate, the most common CFA (Hunt et al., 2005; Stock and Feragen, 2016). In contrast, investigations of psychological adjustment to other rarer CFAs, such as Treacher Collins or craniosynostoses, are scarce and highly needed (Feragen and Stock, 2017), and are lacking nationally and internationally. In addition, treatment for rare CFAs often includes a higher number of medical interventions and more complex surgery than cleft lip and palate, and thus the burden of care involved is potentially more challenging for those affected (Myhre and Agai, 2017).

Centralised team approaches also facilitate the investigation of specialised health professionals’ perceptions, thoughts, and experiences regarding patient communication and the challenges involved in shared decision making, as seen from their perspective. Decisions regarding the treatment of CFAs often need to be based on a multidisciplinary health care approach, since certain treatment choices may impact on other aspects of care (as an example, orthognatic surgery aimed at normalising facial appearance may lead to hypernasal speech). The treatment of CFAs therefore also calls for collaboration and communication between all health care professionals across all involved disciplines. To our knowledge, no studies, have investigated the surgeons’ and other health professionals’ perception of the complex CFA treatment journey, their awareness of central communication skills, and their views or experienced challenges regarding shared decision making.

There is a clear need to conduct research into the experiences and support needs of the CFA population, in order to minimise psychological distress, maximise patient and parent outcomes, and optimise the delivery of healthcare (Feragen and Stock, 2017). In particular, qualitative research, which is able to identify key issues and research priorities according to the perspectives of patients and their families, is of central importance (Nelson, 2009). Such work would have the direct potential to improve existing health services for those affected by a CFA, by integrating the patient perspective into care, and through close collaboration between the patient organisation and the treatment team. In 2016, funding for a pilot study was given to the Centre for Rare Disorders (see project manager’s CV), enabling a preliminary investigation of treatment experiences of parents of children with CFA, as well as adult patients. Results indicate that although participants were satisfied overall with the care they received, some aspects of the consultations were emotionally challenging and disempowering, such as a feeling of being on show and being an object of medical interest (Myhre & Agai, 2017).
In summary, there is a clear need for more patient-centered knowledge about how young people and adults born with a CFA, and their parents, experience the treatment journey they embark upon from the first year of life until adulthood. Long-term and centralised treatment increases the need for and the possibilities of a positive physician-patient relationship. Investigating any challenges of the communication process, both from the patients’ and the health professionals’ perspectives, is of central importance, in order to strengthen shared decision making, especially within a group of patients with complex and demanding treatment pathways. Preliminary findings from the pilot study carried out in 2016, which pointed to some patients experiencing decision regarding treatment decisions as challenging could be addressed by implementing an “advocacy” type of intervention, in order to promote a better understanding of treatment options, increase patient involvement, and provide emotional support. Two qualitative studies demonstrating the potentially strong emotional impact of appearance-altering surgery on some patients with CFA (Beaune et al, 2004; Stavropoulos et al., 2011), call to a better understanding of the psychological consequences of surgery. Knowledge generated from the present project would potentially also inform treatment teams of other patient groups who undergo extensive treatment pathways.

**Specific aims of the current project**

A PhD-based project would contribute to enhanced research competence within an under-researched and important field. The primary aim of this project will be to explore treatment-related experiences from three perspectives: specialised health care professionals, patients born with a CFA, and parents of children with CFA. More specifically, the aims will be to:

- Investigate the emotional impact of appearance-altering surgery on patients’ development of self-perceptions and identity, in order to advance knowledge about adjustment to the treatment-related aspects of a CFA

- Examine potential challenges and strengths in physician-patient communication - from perspective of specialised health care professionals, patients, and parents - in order to promote shared decision making and advance knowledge about the processes involved in treatment-related decisions.

- Explore the usefulness of “decision coaching” and whether an intervention tool such as the PEGASUS could facilitate shared decision making between patients with CFAs and health professionals.

**Methods**

**Participants:**
Participants will include adolescent and adult patients born with a CFA, parents of children born with a CFA, and a sample of specialised health professionals involved in the treatment of CFAs. This approach will allow for the collection of multiple perspectives, and will provide an overview of treatment experiences during the different stages of the treatment pathway.

**Pilot study (preliminary data collection):**
The Norwegian National Unit for Craniofacial Surgery holds a multidisciplinary consultation once a month, offering 10-15 patients and families a short meeting with the multidisciplinary team. From September 2016 to May 2017, 57 eligible participants were offered a consultation at Oslo University Hospital. By the end of May, 32 interviews will have been performed (two
adolescents, 7 adults, and 23 parents). Four participants declined participation (7%), whereas 21 participants (37%) were not reached or have been hesitant regarding participation. Funding from the pilot will allow for interviews to continue until December 2017, hopefully adding 20-30 new interviews to the study. A total of 40-50 interviews are therefore expected as a result of the pilot study. Experiences from the pilot have been used to refine, guide and plan the present PhD application.

**Future plans (if successful PhD-application):**

PhD-funding would secure a full-time researcher into the project, in contrast to part-time involvement during the pilot study, and increase future research competence within this field of work. A large number of interviews is necessary to study the research questions from both the users, parents, and health care workers’ perspectives. The PhD project would be based on a total sample of 70-80 patient/parent interviews (Study 1 and 3), in addition to approximately 15 interviews with health professionals (Study 3), providing an invaluable contribution to international and national research within this field. The large sample will secure large enough sub-samples, so that participants’ experiences may be investigated according to underlying variables, such as specific craniofacial diagnoses, gender or age groups, and investigating experiences of young people, adults, and parents separately, in order to be able to focus on potential specific experiences.

**Study 1:** The first study will focus on patients’ and parent’s experiences of appearance-altering surgery and its perceived impact on self-perceptions, and their thoughts and experiences regarding shared decision-making, from a qualitative perspective. The impact of CFAs on quality of life and self-perceptions will also be explored from a quantitative perspective. The dataset of Study 1 will consist of approximately 65 participants.

**Study 2:** Qualitative interviews of health professionals actively involved in the national treatment service of CFAs (n ± 15). All main involved disciplines will be represented, including surgeons, geneticists, orthodontists, and ophthalmologists. The aim of these interviews will be to explore health professionals’ experiences of challenges and dilemmas involved in their communication with patients with CFA, as well as aspects of communication which currently work well, and whether the PEGASUS intervention tool (see Introduction) could contribute to more satisfactory treatment outcomes. Health professionals will also be interviewed at the end of the intervention trial (see gantt chart), in order to explore their perceptions of potential changes in physician-patient communication in consultations where the PEGASUS was implemented. This part of Study 2 may be included in Study 3.

**Study 3:** The PEGASUS intervention tool will be trialled with approximately 10-15 patients. It was developed in order to increase patient involvement in the treatment decision process. A psychologist helps the patient identify his/her individual goals for surgery and what he/she considers indicative of a successful outcome. These are summarised on a PEGASUS sheet and the patient rates each goal in terms of its importance (Range 0-10). The PEGASUS sheet is then used in the surgical consultation to set shared goals and promote concordance between the patient and surgeon. The surgeon rates the probability of achieving each patient-set goal (Range 0-10), thus facilitating a dialogue about whether expectations are realistic, before final decisions are made. The PEGASUS hence needs to be implemented in consultations where treatment options actually exist, and where patients and physicians need to discuss possibilities, expectations, and potential outcomes. Therefore, only patients identified in
advance by the surgeon as coming to the team to discuss treatment options will be invited to try out the PEGASUS.

**Inclusion/exclusion criteria:**
All patients born with a congenital and rare CFA and/or parents who receive an invitation to attend the multidisciplinary clinic during the project period will be invited to participate. Adolescents aged 12-18 will be asked to participate, with parental consent for those aged 12-16. We aim to interview adolescents and parents separately, unless they express a wish for co-joint interviews. Patients with cognitive levels indicative of difficulties in participating in in-depth interviews will be excluded.

**Design:**
The project will employ a mixed methods design, consisting of both qualitative interviews and quantitative patient-reported questionnaires. Semi-structured, individual interviews and psychometrically sound measures have been chosen in order to investigate two main topics: a) communication and treatment-related aspects and experiences, and b) the perceived impact of appearance-altering surgery on self-perceptions and identity.

**Quantitative component (see Appendix, attached document):** Quantitative measures (Strengths and Difficulties Questionnaire, SDQ, PedsQL, WHOQoL, and Harter’s Self-Perception Scales, SPPA), have been selected for their relevance and good psychometric qualities in similar studies (Stock, Hammond, et al., 2016). These generic instruments will tap different aspects of living with a condition such as a CFA, from the patients’ perspective. Instruments will measure quality of life in parents and adult patients (WHOQOL), in adolescents and proxy (PedsQoL), psychological strengths and difficulties (SDQ, parents and adolescents), and self-perceptions of appearance and social acceptance, among other domains (SPPA, adolescents and adults).

**Qualitative component:** An interview guide, informed by recent review studies (Nelson et al., 2009; 2012; Feragen and Stock, 2017; Stock and Feragen, 2016), and developed for the pilot study will be used for the qualitative part of the study. The interview guide has been informed by relevant literature, clinical experience, and the results of the pilot study. Broad interview topics include parents/patients/professionals’ experiences of multidisciplinary clinics, views on undergoing appearance-altering treatment, satisfaction with the results of treatment, instances of good or difficult communication, and perceptions of shared decision making. Interviews will be recorded, transcribed verbatim, and subjected to thematic analysis, according to the six-step protocol outlined by Braun and Clarke (2006). Interviews will require approximately one hour of each participant’s time. Interviews and analyses will be conducted by the PhD-candidate and reviewed by the research team. Feedback from the analyses will be sent to participants as a check of the credibility of the analyses.

Applying both quantitative and qualitative approaches in the same project (triangulation) is expected to increase the validity of the conclusions. The chosen quantitative measures are currently administered to patients groups attending courses and seminars at the Centre for Rare Disorders and/or have been used in large representative Norwegian samples of young people with or without a medical condition, hence providing national comparison groups.

**Procedure:**
Participants will receive information and an invitation to participate when being offered an appointment for a multidisciplinary follow-up at Oslo University Hospital. After having received information about the project, potential participants will be contacted by the PhD-candidate. Consenting participants will be interviewed shortly after the multidisciplinary consultation. Telephone interviews will also be offered as an option if preferable for the participants, or if they do not have time after the meeting with the treatment team.

Those eligible for being offered a pre-consultation based on the PEGASUS (Study 3) will have a consultation with a clinical psychologist previous to the multidisciplinary consultation. The psychologist will have knowledge about treatment options before meeting the patient, and will also be present during the multidisciplinary consultation. A research interview assessing the perceived usefulness of the PEGASUS will be performed after the multidisciplinary consultation. This interview will be conducted by a different psychologist (or a graduate student in psychology), in order to maximise the patient’s possibilities of expressing his/her true feelings regarding the intervention’s usefulness.

**Schedule for the completion of the project (March 2018 – March 2021)**

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**Ethical perspectives**

Ethical approval was granted for the pilot study (Personvernombudet, OUS), but need to be expanded if funding for a PhD is provided, in order to include a further recruitment of participants, adding health professionals and the plans concerning the intervention study.

When approached for information and written consent, potential participants will be given time to consider what they are being asked to do and will be given the opportunity to ask any questions. Participants will be informed that participation is entirely voluntary, that they can withdraw from the project at any time, that their decision of whether or not to participate in the study will not affect the care they receive, and that they will not be personally identified in any subsequent report or journal article.

The interviews will be carried out by a trained clinical psychologist. Few risks to those involved in the project are anticipated, as was also the experience during the pilot study.
Nonetheless, the topics covered in the interviews may include some emotive issues. Further, some of the interviews may be conducted by telephone, which could potentially reduce the psychologist’s awareness of distress in the participants. Participants will be informed about these two issues prior to giving informed consent. In the unlikely event that a participant should become highly distressed, the interview will be stopped and the participant will be given the opportunity to take a break, or to terminate the interview altogether. If needed, relevant referrals or a consultation with a clinical psychologist at the centre will be suggested.

Relevance for participants and patient involvement

A number of benefits are anticipated for participants (patients, parents, and health care professionals). The pilot study showed that patients and their parents had not had any prior opportunities to speak about their treatment-related experiences and therefore valued the chance to tell their story. In addition, the project will provide the opportunity to feed patients’ and parents’ views back into the healthcare service directly, so that results may potentially guide and improve provision of care for patients born with a CFA. Participant feedback will be discussed and acted upon within the multidisciplinary treatment team, as well as through the patient representative organisation, with a view to improve the service which patients and families receive.

The national treatment team supports the present application. Including health professional’s perspectives into the project will support the implementation of results by providing specific information about their experiences of potential challenges in shared decision making. The study will be run in close collaboration with the patient organisation, Norsk Craniofacial Forening. The project’s Advisory group will include at least three patient representatives (representing adults, young people, and parents), to ensure that research questions are of central importance to patients and their families, and is driven by patient-reported priorities. The project has and will be presented at the patient organisation’s yearly meetings (årsmøte). By regular meetings and dialogue with the organisation, and through the project’s Advisory group, patient representatives will be included in the all phases of the project.

Dissemination and communication of results

Findings will be disseminated in the form of at least three academic publications and presentations at national and international conferences. The findings will be used to inform a dialogue about possible improvements within the multidisciplinary treatment team. Participants will be given a lay summary of the findings directly or through Norsk Craniofacial Forening. The PhD thesis will include three publications:


Paper 2: Specialised health care professionals’ experiences of challenges and strengths in physician-patient communication and shared decision making. To be submitted to the Journal of Health Psychology or similar.
**Paper 3**: Facilitating patients’ preparation for shared decision-making in rare CFAs: Feasibility and acceptability of the PEGASUS intervention. To be submitted to *Psychology, Health, and Medicine* or similar.

Collaborations with the Centre for Appearance Research (University of the West of England) and with psychology graduate students interested in the project is expected to lead to more than three publications in total, if funding is granted.

**International and national collaboration**

The project will be run in collaboration with the Head of Pediatric and Craniofacial Neurosurgery and the Norwegian National Unit for Craniofacial Surgery, Dr Bernt Due-Tønnessen and his team, who is fully supportive of the proposed work.

Dr Ingrid Dundas, Associate Professor, Institute for Clinical psychology, Department of Psychology, University of Bergen, supervised a recent thesis based on the pilot study, and will be one of the secondary supervisors. The proposed project will also be performed in close collaboration with the Centre for Appearance Research (University of the West of England). Dr Nicola M. Stock will be a secondary supervisor on Study 1 and 2, a collaboration that has proved to be highly successful in the past (see attached publication list). Dr Nicole Paraskeva or Professor Diana Harcourt will be secondary supervisors for Study 3 (PEGASUS intervention).

**References**


