

## **Experiences of Daily Life and Oral Rehabilitation in Oligodontia - A Qualitative Study**

Solfrid Sjørgjerd Saltnes<sup>1,2</sup>, Amy Østertun Geirdal<sup>3</sup>, Rønnaug Sæves<sup>2</sup>, Janicke Liaaen Jensen<sup>1</sup>,  
Hilde Nordgarden<sup>2</sup>

<sup>1</sup>Department of Oral Surgery and Oral Medicine, Faculty of Dentistry, University of Oslo, Norway, <sup>2</sup>TAKO-centre Lovisenberg Diaconal Hospital, Oslo, Norway, <sup>3</sup>Faculty of Social Sciences, Oslo and Akershus University College of Applied Sciences, Norway

Corresponding author:

Janicke Liaaen Jensen, professor

Department of Oral Surgery and Oral Medicine

Faculty of Dentistry

University of Oslo,

PO Boks 1109, Blindern,

0317 Oslo, Norway

Mobile: +47- 99725545

e-mail: [jljensen@odont.uio.no](mailto:jljensen@odont.uio.no)

## **Abstract**

**Objective:** Quantitative research indicate increased anxiety and poorer mental health related quality of life (QoL) in individuals with oligodontia (congenitally absence of six or more teeth). The aim of this qualitative study was to complement and explore the individuals' experiences of life and oral rehabilitation, hopefully improving the care for these patients.

**Material and methods:** Twelve participants (6 females, 6 males, aged 21-48) with oligodontia and experiences of comprehensive dental treatments, consented to participate in a semi-structured interview. The questions in the interview guide were based on previous research and clinical experience and included both open (i.e. how is your life?) and specific questions (i.e. do health care personnel know enough about your condition?). The interview transcripts were coded and analysed using a phenomenological method of analysis. The Regional Ethics Committee approved the study.

**Results:** The following themes grew out of the data; "feeling of being different", "the burden of treatment", "shared decision-making", "treatment increases self-esteem" and "use of coping strategies". Psychological distress and reduced QoL seemed to be related to negative aspects of the themes, which covered unacceptable aesthetics, reduced orofacial function, the long-term process of oral rehabilitation, and negative experiences with healthcare services. The informants used problem focused and emotionally focused strategies to meet these challenges. Finalizing oral rehabilitation and shared decision making were positive aspects of the themes.

**Conclusion:** All themes expressed by the participants were of importance for experienced QoL and psychological distress, and should be acknowledged by health care personnel when planning and performing treatment.

**Keywords:** Congenital absence of teeth • Psychological distress • Quality of Life • Coping • Qualitative interview

## **Introduction**

Oligodontia is defined as the congenital absence of six or more permanent teeth, excluding third molars. The condition is associated with a reduction in the size and shape of the teeth, delayed tooth formation, anomalies of the enamel, and disturbances of alveolar growth [1]. Oligodontia can be an isolated trait [1] or be part of a general medical condition, with ectodermal dysplasia (ED) being the most common. In addition to teeth, ED affects other ectodermal tissues like hair, skin, nails, and sweat glands [2, 3]. Our studies indicate that there is an emotional cost associated with living with oligodontia, which is consistent with studies on patients with other chronic disorders [4, 5, 6]. Other researchers have also reported that congenital missing teeth poses significant clinical, functional, aesthetic, and therapeutic challenges [7, 8].

Oral rehabilitation and early treatment are important to avoid physiological and psychological problems because of reduced oral function and unacceptable aesthetics [9, 10]. Thorough treatment planning is important [10], jaw growth should ideally be complete before implants and prosthetic devices are inserted. In addition, the treatment phases must be coordinated and timed correctly to ensure long-term success of treatment [11]. Thus, most individuals with oligodontia have to wait many years before treatment is completed.

Reconstructive surgery has been shown to influence positively psychological factors in another craniofacial condition, Treacher Collins syndrome [12]. To our knowledge, very few qualitative studies to explore how individuals with oligodontia face the challenges associated with the condition have been undertaken. It is generally accepted that quantitative and qualitative studies are complementary [13].

Our former quantitative studies [14, 15] showed that individuals with oligodontia had poorer quality of life and more anxiety than comparable groups. It was therefore of interest to let the individuals with oligodontia put words to their experience, and thereby obtain a broader and enriched understanding on how it is to live with the condition. Such data may contribute to provide a more holistic, coordinated, and multi-professional treatment approach that could be useful for patients as well as professionals.

The aim of the present study was therefore to explore the individuals' experiences of living with and being treated for oligodontia, including a possible explanation for the reported increased levels of anxiety and poorer quality of life.

## **Methods**

### *Design*

A qualitative study design was used to generate knowledge grounded in the respondents' experiences due to living with oligodontia.

### *Participants*

Twenty-two participants had, when enrolled in our previous studies [14, 15] consented to be invited to a qualitative interview. One of them was excluded due to participation in another study. Hence, we contacted 21 individuals by phone, of whom 14 were interested in

participating at this time. All of them then received a formal written invitation and consent form. At this point two individuals withdrew, and the final study group included 12 individuals, six women and six men aged 21 to 48 years (Figure 1). Hence, the study group represents a convenient sample. The respondents all had jobs or were studying.

Figure 1 approximately here

All participants had experience from both public and specialist health care service. They had all completed or were in the last stages of their final dental treatment. Data regarding age, gender, number of missing teeth and self-reported information about clinical signs of ectodermal dysplasia and previous dental treatment is presented in Table 1. The participant's reported number and position of missing teeth were verified clinically and is presented in Table 2. The interviews were conducted at the TAKO-centre in Oslo from February to April, 2016.

Place Table 1 and 2 approximately here

### *Qualitative Interviews and Analysis*

A semi-structured interview guide was developed based on findings from the previously reported quantitative studies [14, 15] and clinical experience. The guide included two main topics with subquestions. The first set of questions addressed how the informants experienced life with oligodontia in general, including a global quality of life question. The second set of questions concerned experience with the health care services, including treatment and

treatment planning, public and specialist health care services, and knowledge among the care professionals. One last question was an opening for the individuals to talk about themes they had thought about, but not addressed previously during the interview. One author (SSS) conducted the interviews, and reflected listening gave opportunities to follow-up the answers and develop depth when needed. The interviewer was well known with the conditions (oligodontia/ectodermal dysplasia), but had only met a few of the informants before. All interviews were recorded and transcribed, and all identifying information was removed during transcription.

### *Data analyses*

The transcripts were analysed and coded manually using Giorgi's descriptive phenomenological method of analysis [16]. This method focuses on descriptions of individual experiences, and emphasises the social phenomena and their importance for the informants. It is a method to identify, condense and abstract the qualitative data and describe themes and patterns across an entire dataset. In accordance with the method, interpretative work was used to categorise the dataset into themes through four steps, including examination of underlying ideas and conceptualisations. During the first step, all interviews were read thoroughly several times in order to get a general sense of the whole dataset and an overall understanding of the given information. In the second step, each interview was attended to individually and different units of meaning were delineated within the data, in order to make the dataset more manageable. In the third step, the units of meaning were transformed from the informants' natural attitudes and expressions to the essence of the respondents' experiences. Each meaning unit originally described in the informants' own words was transformed into suitable expressions without losing the participants' expressed meaning (phenomenological procedure

of free imaginative variation). In the last step, meaning units were transformed into consistent statements of the respondent's experiences. Constituents in the essential structure were checked against the data material to determine whether that was where they originated, which is a structure with the essence of the findings (Giorgi (1975) and Malterud (2001)) [13, 16].

The used quotes are "golden summarized quotes" as described by Giorgi that support the synopsis of the empirical patterns and cannot identify specific participants, rather, the voices of several participants are represented in the different quotes. The method also implies that themes not thought about in the planning phase may be identified.

### **Ethical Considerations**

Ethical approval for the study was obtained from the Regional Committees for Medical and Health Research Ethics (2012/2003).

### **Results**

*Self-reported clinical information.*

Information about self-reported signs of ectodermal dysplasia and previous dental treatment is reported in Table 1. The

Place Table 1 approximately here

*Analysis Results*

The overall impression of the material was that the informants expressed a general feeling of satisfaction at the time of the interviews, although their experiences of living with oligodontia varied. Five important themes relating to their experiences were identified: "*feeling of being different*", "*the burden of treatment*", "*shared decision making*", "*treatment increases self-esteem*", and "*the use of coping strategies*".

***“Feeling of being different”***

The informants explained how they used to have very strong feelings of being different prior to treatment. This was specifically related to the absence of teeth and the amount of time they spent receiving dental treatment. They compared themselves with their peers who visited the dentist once a year at the most. Due to problems with perspiration, some participants were unable to participate in physical activities. Nevertheless, their main physical problem was still related to oral function and appearance.

Several of the informants strongly underscored the importance of “having a nice smile”, and that they never smiled with open lips during their childhood and adolescence, at least not in photographs. They experienced negative comments, which often reduced their self-esteem even more.

*“I heard nasty comments, and that was no fun... I was not first in line when it came to girls, to say the least”.*

The participants’ experiences of having a different appearance and their feelings of being different, highlight the effect of oligodontia on psychological distress, low self-esteem, and reduced QoL and also the challenges presented by this conditions in daily life.

The informants noted that they had worried about being a topic of conversation among peers as well as not being attractive partners, especially before and during dental treatment. It was a common experience that the condition “made a mark on their lives”. However, they seldom talked to others about their situation, and most strived to live an ordinary life.



*“I was bullied a great deal in primary school as well as in secondary school. I chose to keep it to myself and did not tell anyone about my diagnosis” and “I felt insecure when communicating; it was difficult and painful to eat, and it was difficult to pronounce different sounds .... It made me worried, stressed, and nervous”.*

### ***“The burden of treatment”***

Participants described waiting for and receiving treatment as burdens associated with psychological distress. It was challenging to wait for the final treatment, even though they were informed that it could not be done until their jaw growth was complete. Several of the participants had tried to make the dental team initiate treatment early. The treatment itself was also described as a psychological burden due to treatment-related pain, temporary solutions that affected their appearance and oral function, and sometimes a lack of information and co-operation within the health care system. Participants reported that they were not prepared for the pain and the discomfort the treatment caused.

*“The treatment was psychologically challenging and tough; maybe I would do it again, maybe not”.*

Some participants reported that there were a number of “wasted” consultations when their expectations differed from those of the clinician. Several also addressed the lack of knowledge about oligodontia and associated symptoms among clinicians, although they found the clinicians to be very technically skilled. This made the informants feel insecure; they wanted a more holistic approach to their condition.

*“Doctors I have talked to understand the term “ectodermal dysplasia” but not how it affects me”.*

Several informants wished that they had received mental health care to reduce their psychological distress and improve their QoL, especially before and during the treatment process. One experience was described like this:

*“I don’t think anyone ever asked me how I was doing on a psychological level. They were concerned about function, and that is, of course, important”.*

Some of the informants had decided to postpone both education and employment until their dental treatment was finished. Several noted that they had their toughest times during their adolescent years when they were waiting for, or were in the early phases, of treatment.

### ***“Shared decision making”***

Informants experienced “ups and downs” related to meeting the dental team: “Ups” when health care personnel saw the “whole person” and used adequate time for shared decision making, and “downs” when there was less focus and room for shared decision making.

One person stated:

*“Maybe they had a plan, but I never understood what it was”.*

Poor communication and coordination was described, both during treatment planning and during treatment itself. In some cases, this resulted in treatment delays and in concerns about treatment choices. Several informants reported that they did not know who was responsible

for follow-up after treatment. Most understood that future treatment and/or repairs would be necessary, but they chose not to worry about it. However, in retrospect, the individuals in this group described how these factors of uncertainty were associated with reduced self-esteem, more psychological distress, and reduced QoL.

As adults, the participants understood why they had to wait for their final dental treatment and the purpose of the many visits. However, those who had an experience of having received sufficient information and were actively involved in the treatment planning process trusted the clinicians and were more positive about receiving treatment in the future. They reported that specialists spent more resources on them than non-specialists, both in terms of giving information and collaborating with other professionals. This made them feel more satisfied and safe. The feeling is summarised in the following quote:

*“Knowing that you can call the dental care services for questions or necessary treatment any time is of very great importance”.*

When health care personnel saw the informants as useful resources, a safer environment was created. The participants then felt that they were taken more seriously and that they received good care. It also made it easier for them to accept the various treatment challenges and seemed to have positive impact on their psychological distress and QoL.

***“Treatment increases self-esteem”***

To hide missing teeth and less aesthetic temporary solutions, several individuals report that they had used chewing gum to create an illusion of teeth. Others developed the habit of holding a hand in front of the mouth when communicating with other people. Now, after

finishing treatment, almost all of them think they have a nice smile and are satisfied with their appearance. Treatment is described as having positively affected self-esteem, mental health, QoL, and well-being.

*“Now I feel good, I feel like others” and “I am finally done”.*

Dental treatment also positively impacted their capacity to function in daily life, improving their feelings of contentment and QoL. Even though some patients still had chewing problems, they found this acceptable. Whereas they previously had worried about appearance, oral health and oral function, most of them reported that they no longer had worries about losing the temporary solutions, what they could eat, pronunciation and bullying; also, they no longer had pain.

*“The treatment changed everything. Now I can smile, I can eat “normally” ... My teeth are perfect”.*

### ***“Use of coping strategies is important”***

Despite the issues discussed above, most participants accepted the fact that they had oligodontia and the burdens that accompanied this diagnosis. They adapted to reality, which for some included waiting for treatment and postponing their education. *“I am fine”* was a common expression, and *“I take one day at time”*.

Some informants noted that being open about their condition from an early age helped them gain acceptance among their peers.

*“It has been a lot to cope with; it was challenging to be different, but openness helped me”.*

On the other hand, others preferred a different strategy and they chose to reject stressful reality by attempting to keep their condition a secret. This was important to the informants, even though they knew that “everyone” already knew about it. Coping mechanisms related to denial and restraint were also used in their relationships with the opposite sex. They felt unattractive, and they refrained from establishing romantic relationships before and during the dental treatment phase. They held back from such relationships until after their treatment was completed.

*“I felt insecure and did not know what to say to a boyfriend, so I stayed away...”.*

Planning in cooperation with family and with engaged therapists was very important in terms of the efforts of the participants to mitigate the effects of stress related to their conditions. A conscious approach to treatment planning and follow-up were described as having a positive impact on the mental health and well-being. The presence of next of kin, usually parents, at treatment sessions provided emotional support. Having older siblings with the same conditions was also beneficial when the informants could see that their siblings attained satisfactory results after dental treatment.

*“Working as a team with therapists and my next of kin, I knew it would be OK. My siblings prepared me mentally; that helped me a lot... I was lucky to have a family that was prepared for everything”.*

Other informants reported having made the best of their situation even though some, in retrospect, thought that they had misunderstood information from health personnel and clinicians. When they felt different, they told themselves, “*After treatment, I will have the nicest teeth*”. Several took explicit actions, for example taking very good care of their teeth, being very aware of what they could eat and informing clinicians about their condition and the importance of being seen as a whole person. They were physically active and they “carried” toothpicks. All of these strategies are used to remove or circumvent the stressor (Figure 3).

Figure 2 approx. here

## **Discussion**

These qualitative interviews provide a basis for a broader understanding of life with oligodontia and how it may affect mental health and QoL. The findings indicate that individuals with oligodontia experience challenges in everyday life, although they mainly refer to the situation before and during dental treatment. This is in line with earlier studies among individuals with rare disorders [17, 18, 19, 20, 21]. At the same time, the participants reported that they, at an overall level, have satisfying lives, which may mean that they adapt to their situation. The themes “feeling of being different” and “the burden of treatment” represent challenges that may explain the elevated levels of anxiety and poor mental HRQoL reported earlier [14, 15]. The efforts they made to avoid showing their teeth, and the fact that the treatment lasted for years and often was painful, may communicate the essence of the levels of anxiety and poor QoL measured by the quantitative questionnaires Hospital anxiety and depression scale and the Short Form 36 in the previous studies.

Negative attitudes and lack of understanding among healthcare personnel, along with the treatment's duration and pain, also seem to have had a negative psychological impact. While some individuals appreciated good relationships with healthcare personnel, a feeling of not being understood or included in the decision making may have contributed to reduced mental health and QoL [22, 23]. All participants had severe oligodontia, lacking 8-24 teeth, which is rare. Although, the diagnoses itself is clear, many other factors associated with the disorder may be unfamiliar to the dental team. Grut and Kvam [22] state that professionals getting involved in situations that are unknown to them, tend to base their judgement on their personal assumptions, followed by incorrect actions representing a barrier for access to adequate services and assistance for people with rare disorders. However, some informants had more positive experiences as they described being well treated, listened to, taken seriously, and given the information they needed. It therefore seems like a holistic approach is important for maintaining good mental health and good QoL. This is much in line with a study by Huyard (2009) in which informants (both patients and parents) were interviewed regarding their experiences of living with a rare disorder [24]. In fact, these informants underscored that the rare disease itself was not the most important cause of their specific difficulties, but rather the failure of health professionals to meet their specific needs. When health professionals behaved in a way that satisfied the moral needs of the patients and their relatives, their experiences were rather positive, and conversely, negative when their needs were unmet [24]. Shared decision-making was an important aspect in this regard. In fact, individuals with chronic conditions often know more about their condition and its management than the professionals, and their knowledge may contribute to adequate treatment and follow-up, as also underscored by Petersen [25].

Our findings show that the participants used coping strategies when meeting everyday challenges and in reducing the burden of their condition. Previous studies have also evaluated

various coping strategies used to manage their condition by individuals with rare disorders. Such factors include to act, try to be like everyone else, and try to live a normal life, as far as possible [18, 19, 21, 23, 26, 27] . For example, many informants in the present study accepted that oligodontia made them different from their peers, and they adapted to this reality. Planning helped them confront the stressors associated with oligodontia, and they articulated their desire to grow as people and to make the best out of their situation as well as to take steps to remove the stressors. All of these represent active coping. In line with earlier research [28], our results indicated a relationship between the individuals' experiences of good mental health, QoL, and coping. Shared decision making, as well as the importance of completion of treatment, the experience of self-esteem and the use of coping strategies were all reported as positive factors. As described above former research showed that oral rehabilitation and early treatment were important to avoid physiological and psychological challenges [9, 10], and our findings are in line with this. Another positive finding was that instead of using coping strategies with emotional focus, for example denial, the participants mainly reported acceptance and adaption to the situation. The themes "Feeling of being different" and "the burden of treatment", in addition to lack of shared decision-making seem, on the other hand, negatively impacted on mental health and QoL in this group.

Oligodontia is a complex condition that poses challenges in many aspects of life. The condition itself, or its consequences, can cause individuals to experience psychological distress and poor QoL. There may be a discrepancy between expectations and real life. However, the participants did not seem to be trapped in psychological distress. Instead, they said that overall, they think they have a satisfying life. This is consistent with our clinical experience. The elevated anxiety level and reduced QoL [14, 15] may be related to the feeling of uncertainty about the future, but these individuals seemed to manage their psychological burden by using coping strategies relevant to their situation. This study cannot indicate what



coping strategies are the best to limit the psychological distress. More research to investigate and answer such questions must be done in the future.

However, the experiences and thoughts expressed by the participants in the present study clearly indicate that mental aspects should be evaluated in individuals with oligodontia. To address this issue, more interdisciplinary research and clinical work should be performed. A multidisciplinary approach with experienced professionals who are well acquainted with the diagnosis, may contribute to more detailed information about the diagnosis, treatment planning and treatment alternatives. It may be beneficial for participants to be given greater access to information about network groups and support groups. Placing more focus on patient participation in treatment planning and better preparation before treatment would probably contribute to decreasing uncertainty and preventing unnecessary psychological distress. In addition, the professional dental team should strive at heightening their communication and shared decision making skills.

One strength of this study was that the interview guide was based on our previous research and long-term clinical experiences. The open-end questions provided details about the individuals' experiences of living with and being treated for oligodontia. In order to address the questions we have identified through experience and research, a semi-structured approach was chosen. A semi-structured interview is open, allowing new ideas to be brought up during the interview as a result of what the respondent says. However, it also ensures that all the themes the researcher wants to be covered during the interview are addressed.

The respondents had experiences from both public and specialist health care services. A few participants had not finalized their final treatment, and the sample differed in type of treatment received as well as in age. This may be seen as a limitation in the material. However, it may also be seen as a strength as diversity within the material may add a broader understanding of how life with and treatment of oligodontia actually is. As the same person

performed all interviews and as she had knowledge about the conditions and experience in oral healthcare, she was able to extract information important for the practical organisation of future oral healthcare for this patient group. On the other hand, this could be a limitation. The interviewer could be suggestive in the interviews and have a mind-set of her own that would limit the understanding of the informants. It was therefore of importance to have a conscious attitude to this topic throughout the interviews.

Another limitation of the study is that the qualitative method is context-specific and cannot be generalised to the overall patient group. Nevertheless, the chosen method is valid and provides insights into the experiences, attitudes, and opinions of this group. Despite that the group of respondents was not a strategic selection, but a convenient one, and that the low number of participants can be seen as a limitation, saturation was reached. During the last interviews, no new topics came up, only minor variations in the answers to the topics asked for.

A question might be what coping strategies are the best for the outcomes. Due to the qualitative method it is not possible to tell which strategies are best in this regard.

In conclusion, psychological distress and reduced QoL seem to be related to unacceptable aesthetics, reduced orofacial function, the long-term process of oral rehabilitation, and experiences with healthcare services. The informants used a range of coping strategies that seemed important for meeting everyday challenges. Although coping strategies were important, oral rehabilitation itself, as well as shared decision making, was crucial. It seems important to put greater emphasis on the psychological challenges facing these patients and to encourage their use of coping strategies in order to provide a more holistic, coordinated, and multi-professional treatment approach.

**Acknowledgments**

We thank all of the participants for their contributions to this study.

**Conflicts of Interest**

The authors declare that there were no conflicts of interest regarding the conduct of this study.

**Informed Consent**

All procedures followed were in accordance with the ethical standards of the responsible committees on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all participants before being included in the study.

## References

- [1] Schalk-van der Weide Y. Oligodontia. A clinical, radiographic and genetic evaluation. [PhD thesis] Utrecht: University of Utrecht; 1992. 1992.
- [2] Freire-Maia N. Ectodermal dysplasias. *Hum Hered.* 1971;21:309-312.
- [3] Freire-Maia N. Ectodermal dysplasias revisited. *Acta Genet Med Gemellol (Roma).* 1977;26:121-131.
- [4] Pasculli G, Resta F, Guastamacchia E, et al. Health-related quality of life in a rare disease: hereditary hemorrhagic telangiectasia (HHT) or Rendu-Osler-Weber disease. *Quality of life research : an international journal of quality of life aspects of treatment, care and rehabilitation.* 2004;13:1715-1723.
- [5] Geirdal AO, Dheyauldeen S, Bachmann-Harildstad G, et al. Quality of life in patients with hereditary hemorrhagic telangiectasia in Norway: a population based study. *Am J Med Genet A.* 2012;158a:1269-1278.
- [6] Velvin G, Bathen T, Rand-Hendriksen S, et al. Work participation in adults with Marfan syndrome: Demographic characteristics, MFS related health symptoms, chronic pain, and fatigue. *Am J Med Genet A.* 2015;167a:3082-3090.
- [7] Al-Ani AH, Antoun JS, Thomson WM, et al. Hypodontia: An Update on Its Etiology, Classification, and Clinical Management. 2017;2017:9378325.
- [8] Locker D, Jokovic A, Prakash P, et al. Oral health-related quality of life of children with oligodontia. *International journal of paediatric dentistry.* 2010;20:8-14.
- [9] More CB, Bhavsar K, Joshi J, et al. Hereditary ectodermal dysplasia: A retrospective study. *J Nat Sci Biol Med.* 2013;4:445-450.
- [10] Sclar AG, Kannikal J, Ferreira CF, et al. Treatment planning and surgical considerations in implant therapy for patients with agenesis, oligodontia, and ectodermal dysplasia: review and case presentation. *J Oral Maxillofac Surg.* 2009;67:2-12.
- [11] Papez J, Dostalova T, Kriz P, et al. Dental implants and skeleton maturation as factors influenced implant insertion by adolescent patients with missing permanent teeth: 5-year prospective study. *IOSR Journal of Dental and Medical Sciences (IOSR-JDMS) Volume 14, Issue 9 Ver VIII PP 43-48.* 2015.
- [12] Arndt EM TF, Lefebvre A., Munro I. R. . Psychosocial adjustment of 20 patients with Treacher Collins syndrome before and after reconstructive surgery. *British Journal of Plastic Surgery,* 40, 605–609. 1987.
- [13] Malterud K. Qualitative research: standards, challenges, and guidelines. *Lancet (London, England).* 2001;358:483-488.
- [14] Geirdal AO, Saltnes SS, Storhaug K, et al. Living with orofacial conditions: psychological distress and quality of life in adults affected with Treacher Collins syndrome, cherubism, or oligodontia/ectodermal dysplasia-a comparative study. *Quality of life research : an international journal of quality of life aspects of treatment, care and rehabilitation.* 2015;24:927-935.
- [15] Saltnes SS, Jensen JL, Saeves R, et al. Associations between ectodermal dysplasia, psychological distress and quality of life in a group of adults with oligodontia. *Acta odontologica Scandinavica.* 2017;75:564-572.
- [16] Giorgi A. An application of phenomenological method in psychology. In: Giorgi A, editor. *Duquesne studies in phenomenological psychology Vol II.* Pittsburgh: Duquesne University Press; 1975.
- [17] Barlow JH, Stapley J, Ellard DR. Living with haemophilia and von Willebrand's: a descriptive qualitative study. *Patient Educ Couns.* 2007;68:235-242.

- [18] Smith N, Bartholomew C, Jackson S. Issues in the ageing individual with haemophilia and other inherited bleeding disorders: understanding and responding to the patients' perspective. *Haemophilia : the official journal of the World Federation of Hemophilia*. 2014;20:e1-6.
- [19] Palareti L, Poti S, Cassis F, et al. Shared topics on the experience of people with haemophilia living in the UK and the USA and the influence of individual and contextual variables: Results from the HERO qualitative study. *Int J Qual Stud Health Well-being*. 2015;10:28915.
- [20] von der Lippe C, Diesen PS, Feragen KB. Living with a rare disorder: a systematic review of the qualitative literature. *Mol Genet Genomic Med*. 2017;5:758-773.
- [21] von der Lippe C, Frich JC, Harris A, et al. Experiences of Being Heterozygous for Fabry Disease: a Qualitative Study. *J Genet Couns*. 2016;25:1085-1092.
- [22] Grut L, Kvam MH. Facing ignorance: people with rare disorders and their experiences with public health and welfare services. *Scandinavian Journal of Disability Research*. 2013;15:20-32.
- [23] Garrino L, Picco E, Finiguerra I, et al. Living with and treating rare diseases: experiences of patients and professional health care providers. *Qual Health Res*. 2015;25:636-651.
- [24] Huyard C. What, if anything, is specific about having a rare disorder? Patients' judgements on being ill and being rare. *Health Expect*. 2009;12:361-370.
- [25] Petersen A. The best experts: the narratives of those who have a genetic condition. *Soc Sci Med*. 2006;63:32-42.
- [26] Jaeger G, Rojvik A, Berglund B. Participation in society for people with a rare diagnosis. *Disabil Health J*. 2015;8:44-50.
- [27] Dures E, Morris M, Gleeson K, et al. The psychosocial impact of epidermolysis bullosa. *Qual Health Res*. 2011;21:771-782.
- [28] Nipp RD, El-Jawahri A, Fishbein JN, et al. The relationship between coping strategies, quality of life, and mood in patients with incurable cancer. *Cancer*. 2016;122:2110-2116.