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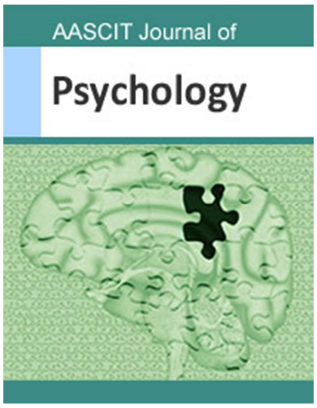

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Ectodermal Dysplasia – A Patient Psycho-Social Perspective

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Abstract

Introduction: This retrospective study investigated the psycho-social experience of 20 year old patients who were born with ectodermal dysplasia and have undergone many years of multidisciplinary treatment. In particular, this study sought to define the limitations of having ED, the impacting personal issues and perceptions of care. The advantage of this cohort was that subjects have had time for reflection and life experience following their treatment. *Materials and Methods:* This study utilised two collection instruments - a condition-specific questionnaire designed to obtain both qualitative and quantitative responses and a follow-up qualitative questionnaire for those who indicated that their lives had been greatly impacted by ED. Participants were 18 years of age and over and had completed their treatment milestones. Participants were contacted by telephone prior to the self-report questionnaires being forwarded by post. *Results:* Overall, respondents were most impacted by issues relating to: self-esteem, attitude to life, independence, special people friendships and future plans. The most significant psycho-social impact of having ED is the degree of influence on respondents' self-esteem. In particular three respondents (42%) reported that having ED had dramatically affected their self-esteem and the course of their lives as a consequence. *Conclusion:* Although there were small participant numbers in this study, it provided a significant proportion that reported having been adversely affected as a result of having this condition. The degree to which two of the respondents were affected by ED requires further investigation and consideration in regard to how this may be identified for future patients, what guidelines are required for offering assistance, and what services are available to help address these issues.

1. Introduction

Ectodermal dysplasia (ED) is a heterogeneous group of inherited disorders characterised by developmental abnormalities manifesting in at least two different ectodermally derived systems [1]. They affect approximately one in every 100 000 live births [2]. The types of EDs are recognized by the combination of physical features that an affected person has and the way in which they are inherited. They may include defects of the hair, nails, teeth, and sweat glands. Other symptoms may include absent or decreased lacrimal production, decreased skin pigment, offensive nasal discharge, heat intolerance, inability to perspire, large forehead, congenital tooth disorders, low nasal bridge, poor hearing, impaired temperature regulation, poor vision, sparse hair and fine, flaky skin [2-4]. It has been previously suggested that patients with ED may also have intellectual disability [5-6]. Tooth disorders and deficiencies associated with ED may require complex multi-staged treatment. Dental evaluation and treatment is recommended at an early age to determine whether prosthodontics may be suitable as part of the patient's dental management and to assist with the psychosocial impact, in particular teeth and facial appearance [7-11].

The impetus for this study was the result of an enquiry made to the Dental Department from a past patient who wished to be put in contact with support groups or other individuals with this condition.

This retrospective study investigated the psycho-social experience of 18 to 20 year old patients who were born with ectodermal dysplasia and have undergone many years of multidisciplinary treatment. In particular, this study sought to define the limitations of having ED, the impacting personal issues and perceptions of care.

Aims: The aims of this study were to:

- obtain an understanding of the patient experience of ED;
- examine the individual's perception of their treatment & determine how these results may influence and improve future services;
- determine if there is a likely pattern of psycho-social needs and the availability of appropriate services and solutions to assist in dealing with those needs.

2. Materials and Methods

This study utilised two collection instruments - a condition-specific questionnaire designed to obtain both qualitative and quantitative responses and a follow-up of further comments and reflections for those who indicated that their lives had been greatly impacted by ED. Participants of 18 years of age and over who had completed their treatment milestones were requested for enlistment. The advantage of this cohort was that subjects have had time for reflection and life experience following their treatment. This offered a pool of 12 participants. Three could not be contacted and two refused to participate leaving a response rate of 7 from 9 (78%). Descriptive statistics were calculated using Microsoft Access and Microsoft Excel.

Participants were contacted by telephone prior to the self-reporting questionnaires being forwarded by post. The questionnaire was comprised of questions from areas of demographics, impact of having ED, attitude to life and satisfaction with service at PMH.

Four of the seven respondents were male, 5 were Caucasian and 2 were of Asian racial heritage. Five respondents had Hypohidrotic form of ED, 1 had Ectrodactyly Ectodermal Dysplasia (EEC) and 1 had Rapp Hodgkin syndrome or Anhidrotic ED (and was the only participant who also had a cleft palate.) All participants had resided with, and were raised by their birth parents. One of the respondents had divorced parents. Five participants resided in the metropolitan area and two in country areas. The three female respondents all reported having family members with ED. Two reported that their mother also had ED and one reported that their father had ED. No other family members were reported as being affected.

Four reported severe effects of ED to their teeth (3 moderate), two to their hair (2 moderate, 3 minor), one to their skin (2 moderate), one to their sweat glands (2 moderate) and one to their nails (1 moderate). One reported moderate effects of chest infections and one reported severe effects of skin/nail infections (1 moderate, 1 minor). The average age was 22 years.

3. Results

Overall, respondents were most impacted by issues relating to: self-esteem, attitude to life, independence, special people friendships and future plans (Figure 1).

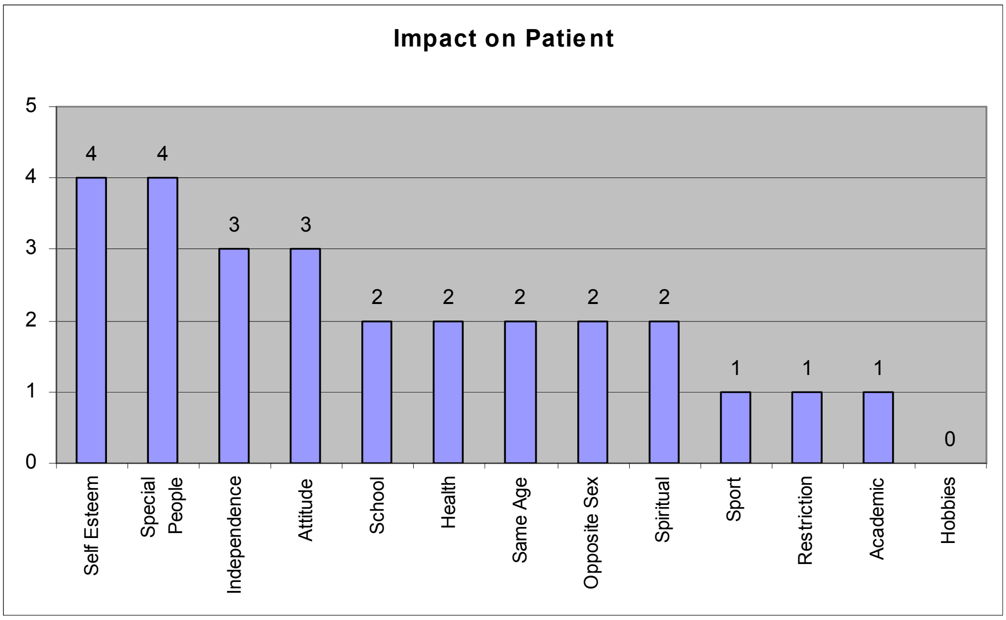

Figure 1. *Impact on life.*

4. Impact on Life

The most significant impact was reported to be self-esteem with 4 (57%) reporting that ED had impacted their self-esteem. In particular three respondents (42%) reported that having ED had dramatically affected their self-esteem and the course of their lives as a consequence.

Interestingly, of the three respondents who reported no effect on their self-esteem, one reported having been severely affected in regard to their physical appearance (hair, teeth and skin). Of the four respondents who reported that ED had affected their self-esteem, two reported only moderate and minor effects to their physical appearance (teeth and hair).

Four of the respondents reported having difficulty meeting/attracting a special person for a relationship. Three reported that ED had an impact on their independence, this was in terms of being tied to appointments and two reported not having the confidence to be in the world. The same three reported the ED impacted their attitude to life, especially in terms of feeling self-pity and a lack of self-confidence.

5. Teasing

Four (57%) of the respondents reported having been teased for reasons due to their ED condition. Of these, two (29%) also reported being teased for other reasons, but still reasons related to their physical appearance. Those who reported no teasing were those with mild/minor ED manifestation in their physical appearance.

6. Support from Others

When asked from whom they rated the importance of support, respondents reported their mother, treatment givers, employer and their father as the most important. However treatment givers, employer, teachers, friends and peers did not meet the level of support required.

7. Appearance of Teeth

Respondents were asked to rate both the importance and satisfaction of their teeth appearance (Figure 2). Four (57%) rated the appearance of their teeth as extremely important but only 2 reported their satisfaction with the appearance of their teeth as ‘extremely’ satisfied. Two (28%) reported ‘not very’ and one rated for ‘very’ satisfied grouping. Two rated the importance of their teeth appearance as ‘very’ and rated their satisfaction as ‘extremely’ (14%) and ‘very’ (14%). The 2 who rated the importance of the appearance of their teeth as ‘somewhat’ rated their satisfaction as ‘extremely’.

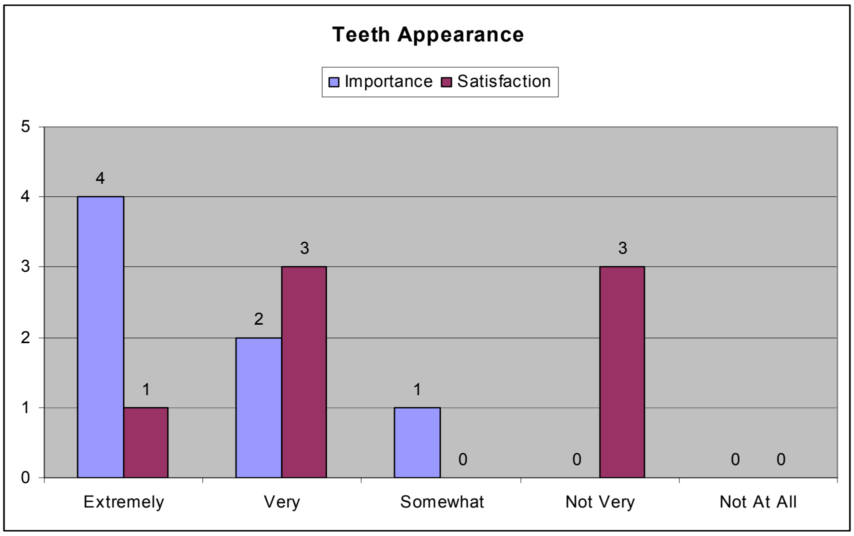

Figure 2. *Appearance of teeth.*

8. Impact on Patient

Respondents were requested to comment on the effects of ED on certain parts of their lives (Figure 3). Six (86%) reported that ED had affected their attitude to life. Two reported that it had made them more understanding toward others and one reported that it has made them wary of medical treatment and personnel. Three reported that ED is the cause of their low self-esteem (two described having experienced self-pity) and depression/anxiety issues. Of these, two reported being diagnosed with clinical depression and perceived their lives as having been ‘hard’ or ‘unbearable’ at times. The same two also reported to have ‘wasted’ a considerable amount of their lives, one becoming alcohol and drug dependant and the other spending many years not emotionally able to participate in society. In contrast, one respondent with severe physical manifestations reported that they reached a point in their lives where they just refused to allow ED to continue to affect their life. Two respondents reported that having their own children had a major positive impact on both their attitude to and enjoyment of life.

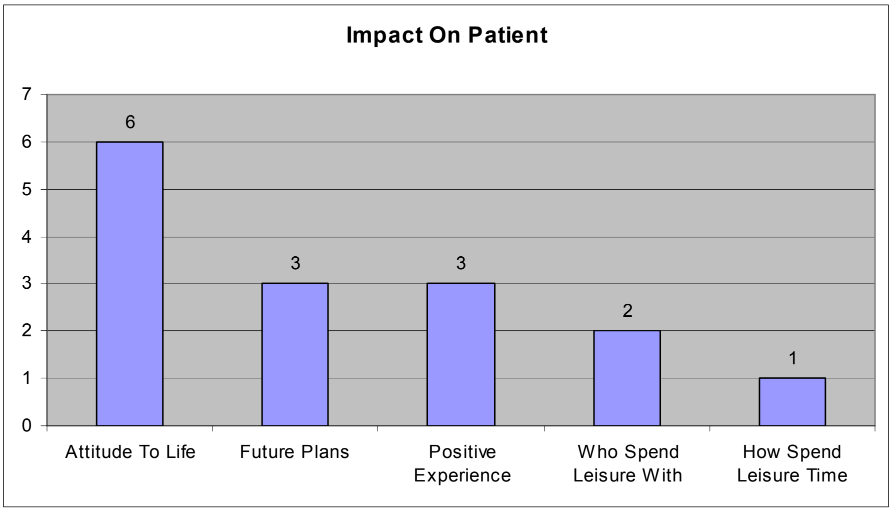

Figure 3. *Impact on patient.*

Three respondents reported that ED had affected their future plans and this involved the many appointments which restricted travel plans and two (28%) reported their concern of having children should they pass on the condition. Of the two respondents that have had children one has passed the condition on to their child.

Of the 7 respondents, only two felt that they would have benefited from talking to a counsellor, support group or others with ED. One felt they would have liked to talk with the clinic co-ordinator to be able to have a clear and simple understanding of the treatment process.

All respondents felt adequately prepared for treatment. Six (86%) respondents reported that making their own care decisions was important to them and 5 reported that they had been given that opportunity.

9. Limitations

The small patient numbers prevent this study from being statistically significant.

10. Discussion

A person's response to life's experiences is always subjective and may be awarded varying importance dependent upon many life-impacting factors, and may be reflective of the outcomes and responses received from their previous experience. Minor incidents or moderate physical defects may have major importance to one individual and yet may be of minor importance to another or to the same individual at another time in their life. Although it may be reasonable to assume that those patients with the most severe manifestations of a condition in their physical appearance would suffer the greatest impact from factors such as teasing, low self-esteem and self-confidence, and poor attitude to life, as shown with the results from this study, it may not necessarily be the reality. Even though the experiences and responses from this study occurred many years before, and may no longer be current, they may still act as a timely reminder of the potential psycho-social issues of all ED patients. As with all health service provision, all members of that particular unit or institution would like the patient to receive the best treatment experience possible.

However, defining where the responsibility lies for a patient to be offered counselling services requires consideration. Practitioners need to have the confidence in their ability to engage with the patient and to be able to know when it is appropriate to refer the patient for assessment and counselling. In order to offer such services outside of standard protocols a practitioner would need to have an established current treatment relationship with the patient and preferably also with their parent in order to identify the need. As most of the treatment occurs for a short amount of time and for many with long intervals between appointments, this may not be possible. Equally, without the established treatment relationship, it may not be possible for either the patient or the parent to feel the confidence or comfort to request referral for counselling services from the practitioner. Practitioner-level psycho-social training may be beneficial in providing a knowledge base for referral for at-risk patients.

11. Conclusion

Although there were small participant numbers in this study, it provided a significant proportion that reported having been adversely affected as a result of having this condition. Respondents were most impacted by issues relating to: self-esteem, attitude to life, independence, special people friendships and future plans. The most significant psycho-social impact of having ED is the degree of influence on the individual's self-esteem and their attitude to life. The degree to which two of the respondents were affected by ED requires further investigation and consideration in regard to how this may be identified for future patients, what guidelines are required for offering assistance, and what services are available to help address these issues. The degree to which individuals require assistance is not dependent upon the severity of their condition – but upon their personal response to their condition. This study has indicated that those with relatively minor physical effects may suffer from psychosocial issues which require referral just as much as those with more severe physical effects. Practitioners need to be aware and vigilant of this and refer when required.

Recommendations

Evaluate the potential need for:

- additional counselling services for patients with ED;
- practitioner-level psycho-social training for those treating patients with ED;
- providing literature which may assist patients to deal with the potential negative response from others including teasing;
- providing literature which offers the contact details of support groups.

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