

PSYCHOSOCIAL PRESSURES OF HAVING BETA-THALASSEMIA

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Abstract

Having a chronic disease presents a series of lifelong problems for the patient and the family. In regards to beta-thalassemia major (BTM), the patient has to worry about the effects of medical care on their body from such a young age. However, after interviewing Patient S, it has been stated that maintaining his mental health was the most difficult part of having BTM. Although some patients can adjust to their lives, not everyone has the same story in regards to the way they view themselves and the way they interact with others. The goal of this literature review is to address the psychosocial pressures that Patient S discussed with a focus on engaging in romantic relationships. This requires exploration of the psychological and physical development of the patient with the disease which will give a better picture of the sociological development that Patient S explained. There are several studies of BTM that took place in western countries where beta-thalassemia is not as prevalent and with medicine that is of a different standard. However, due to Patient S being from a country that has a higher prevalence of BTM with less of these resources, it was only appropriate to include studies representing this population.

Keywords: *Psychology, Sociology, chronic conditions.*

1. Introduction

Having a chronic disease presents a series of lifelong problems for the patient and the family. In regards to beta-thalassemia major (BTM), the patient has to worry about the effects of the medical care on their body from such a young age. However, receiving medical care seems to be the easiest part of the process. After interviewing Patient S, it has been stated that maintaining his mental health was the most difficult part of having BTM. Although some patients can adjust to their lives, not everyone has the same story in regards to the way they view themselves and the way they interact with others.

Beta-thalassemia was first identified in 1925 by Dr. Thomas Cooley when he discovered the similarities in the changes of certain patient's bones, as well as their clinical histories (Rich, 1956). Due to the lack of knowledge of the different types of anemia and the fact that this anemia presented differently than the previously discovered ones, this new disease was called Cooley's anemia. The discovery of this disease led to various genetic studies that highlight the inheritance pattern of this disease, as well as the populations that are more prone to it.

Discovering a chronic disease such as BTM can lead to medical advancements that could help patients with the disease live a better life, or could help eradicate the disease in certain instances. Eradication of BTM would be through pre-marital screening programs that are put in place in highly endemic areas to hinder higher-risk marriages (Hashamizadeh and Noori, 2013; Zeinalian et. al., 2013). These programs have become more prevalent amongst societies as time has progressed but were not as popular about 50 years ago when Patient S was born. These programs have helped reduce the number of BTM births among endemic populations, but the couple has to be willing to get tested before marriage (Waheed et. al., 2016). In some cases, couples did not get tested for BTM and ended up having a child diagnosed with this disease which led to debilitating consequences for the family. In Patient S's case, his parents were not tested before marriage. In instances when patients are born with BTM, there are medical advances that can provide the patients with a better life. Currently, patients with BTM are required to get blood transfusions ever so often to attempt to correct the anemia (Galanello and Origa, 2010). The repeated blood transfusions require iron overload therapy to avoid various complications that come with their lifelong therapy. Although patients can reduce certain complications that are prevalent in iron-overload, other complications come with the treatment such as delayed or arrested development which puts the patients behind their peers physically and mentally.

The discovery of a chronic disease can also lead to stigmatization which is due to the ignorance of the community about a patient's condition. Awareness of BTM and what it indicates is strongly correlated with those of a higher socioeconomic status than those who live in more remote areas (Wong et. al., 2011). Public awareness has increased in recent years, but there are still populations who are unaware of the disease which leads to problems in addressing those affected by it. Due to Patient S being born over 40 years ago when the disease was considered a death sentence in the Mediterranean, he was a victim of stigmatization that followed him when he applied for work or even tried to find a life partner.

The goal of this literature review is to address the psychosocial pressures that Patient S discussed with a focus on engaging in romantic relationships. This requires exploration of the psychological and physical development of the patient with the disease which will give a better picture of the sociological development that Patient S explained.

2. Literature review

BTM is a chronic disease that is debilitating to the patient and the family. Due to life-long treatment, patients with BTM develop differently in regards to their physical appearance and their psychological processes. Due to the differences that BTM patients feel in comparison to their peers in regards to their physical and mental health, this leads to sociological problems when interacting with others. Patient S spent a lot of time discussing the difficulties he and his friends with the disease had in regards to finding a partner due to the stigmatization around the disease, the developmental delays the disease causes and what it could mean for their future offspring. In this literature review, I will discuss the psychological and physiological development of patients with BTM which can lead to sociological dysfunction focusing on issues in relationship development through various relevant papers based on the information received from Patient S during our interview.

2.1. Psychological

The current literature on the topic of the psychological pressures of having a chronic disease such as BTM has been heavily explored. Having a chronic disease is a mind and body game. The patient must be able to handle the treatments involved for their entire lives and the patient must also be able to mentally deal with the psychological burden a chronic disease brings.

Many BTM patients feel the effects of the disease on their bodies which could be the result of their developmental delay and not being able to participate in certain activities like their peers. Inability to participate in these activities could reduce the patient's self-esteem and self-efficacy since they do not feel like they are the same as others around them. Patient S discussed how he enjoyed playing soccer when he was younger, but he had restrictions that other children did not have due to his disease. That dream of becoming a famous soccer player that many young children have was not even a dream since he knew his condition would not allow that to happen. However, he was satisfied with the way he grew up because many people he knew with the disease had more restricted lifestyles so they did not have the chance to experience certain pleasures that come with being a kid.

Not being able to participate in various activities or not having a similar appearance to your friends can lead to social isolation. Due to the societal rejection that certain patients feel depending on their region and how old they are, depression is prevalent amongst patients with BTM (Koutelekos and Haliasos, 2013). Patient S spoke about how his life is a roller coaster of emotions. Sometimes he feels a lot better due to his treatment, but other times when he feels down, he wants to just stop his treatment. He followed this by saying that he knew if he stopped his treatment, then he is signing his death warrant so that was not an option. But he has been around many people with BTM who do not want to live anymore so their treatment compliance plummets and they end up passing away. Patient S did not disclose having a psychological disorder, but he did acknowledge bouts with depression which is common amongst patients with BTM as mentioned earlier. Although diagnosable mental illness was not the case for Patient S, but feeling the effects of social isolation and could lead to other psychological disorders "to name a few [they] can be referred to [as] somatization disorders (SOM), depression especially major depression disorder (MDD), obsessive-compulsive disorder (OCT), tic disorders, oppositional defiant disorders (ODD), psychosis, nocturnal enuresis nocturnal. In addition, somatic complaints, and interpersonal sensitivity" (Naderi et. al., 2012). Patients with BTM are more vulnerable to psychological disorders, especially if they lack primary and secondary social support (Naderi et. al., 2012). Due to the complexity of some of these psychological disorders, medical compliance will decline. A more popular phenomenon for BTM patients is psychiatric help due to them being at high risk (Hakeem et. al., 2018). Lifelong therapy is suggested due to BTM being a lifelong disease.

2.2. Physical

Due to the treatment involved with this chronic condition, patients do not develop at the same speed as their classmates. It is quite common in the literature for patients with BTM to have delayed or even arrested puberty (Kyriakou and Skordis, 2009).

Although Patient S is male, some women experience issues due to BTM in regards to consonance during puberty (Al-Rimawi et. al., 2005). Female patients tend to be of smaller stature and appear less developed than their classmates without the disease. When the patients were finally going through puberty, they had to later worry about their fertility if they wanted to give birth (Roussou et. al., 2013). Women who wanted to have children were able to have children through fertility treatments. However, having children naturally with a partner is difficult. Alternative reproductive techniques are usually utilized in patients BTM (Miri-Aliabad et. al., 2015). This finding is confirmed in two additional literature pieces that discuss how the oxidative stress of BTM treatment leads to overproduced circulating free radicals leading to increased infertility (Roussou et. al., 2013; Moayeri and Oloomi et. al., 2006).

In males, there are similarities to females in regards to their physical developmental delays. Males have a shorter stature and their features may not appear as mature as their friends due to the hypogonadism that is a common side effect of BTM treatment (Galati, 2003). Patient S revealed that he was shorter than many of his classmates, but he was not sure why. Upon research, it is evident that hypogonadism is most likely the cause. Due to treatment, boys can commence puberty like their friends, but then they have other issues to think about later in life. Erectile dysfunction and azoospermia are common side effects of the recurrent treatment (Giammusso et. al., 2000). These conditions were confirmed in another study which shows that free radicals and iron deposition in the pituitary glands and the testes lead to problems with fertility (De Sanctis et. al., 2012). There are some advances in male infertility such as providing the patients with antioxidants, but the studies are in their infancy (Elsedfy et. al., 2018).

2.3. Sociological

Based on the side effects and complications BTM described above, these patients tend to have difficulty in social functioning. As discussed before, many patients with BTM have difficulties participating in certain activities as their unaffected friends which could lead to social depression and reduced medical compliance (Koutelekos and Haliasos, 2013). Feeling as though a person is different from everyone else can mold them the rest of their lives. Due to the advances in BTM treatment, patients can live much longer than before which allows them to get to reproductive age. Patient S knew he was different from a lot of the people around him, but when he got to his adolescent and young adult years when he was looking for a partnership was when it became more evident.

As mentioned before, patients with BTM have developmental delays due to their treatment which results in delayed or even arrested puberty. One of the most visible side effects of having BTM is being much shorter than your classmates (Galati, 2003). Upon meeting Patient S, he had a small stature which he later explained that he was a lot smaller than his classmates due to his treatment. Height is not a problem in most relationships, but Patient S had to worry about other issues that his counterparts did not. Patient S wanted the closeness and intimacy that came with having a partner and being married, but he also had to think about whether he would be rejected and then whether he would be able to bear offspring. A finding suggests this is a common thought amongst patients with BTM since “one serious difficulty of social integration, deriving mainly from the lack of a normal sexual function: often prevents to construct[ion of] a family” (Messina et. al., 2018). Patient S did not go into detail about his gonadal functioning, but as previously mentioned, males with BTM were found to have erectile dysfunction and infertility. These factors lead to patients having to approach relationships differently than their counterparts. Although Patient S was able to get married and have two children with his wife, he is an outlier in the data. The few studies focusing on the patients in regards to social relationships found that patients do not usually seek marriage and even fewer want to have children (Haghpanah et. al., 2013; Messina et. al., 2008).

3. Discussion

While preparing for this literature review, I became more informed about Patient S’s condition regarding how this chronic disease can cause psychological and physical differences which can adjust the way a patient interacts with others around him. Although there was a lot of research in certain areas of BTM, there are some areas that are lacking investigation which leaves a few unanswered questions. In this discussion, I will pose some of the downfalls present in this literature review.

3.1. Psychological

While researching the mental health of patients with BTM, there was a lot of mention of psychological disorders that patients with BTM are susceptible to especially if they have co-morbidities (Hakeem et. al., 2018; Messina et. al., 2008; Naderi et.al., 2012). Although patients with BTM are more likely to get mental illnesses, not all of them do. There was not a lot of research about patients without mental illnesses. In regards to Patient S, he did not disclose having a mental illness, but he did reveal that he has a rollercoaster of emotions sometimes due to his illness. He also revealed how his family was very supportive of him so he had more social support than others. Lack of social support is a predictor of developing a mental disorder that puts a strain on the patient which can change the course of their life (Naderi et. al., 2012). This is important in regards to my paper because if patients with BTM who do not have mental disorders can make a smoother adjustment into society and have better sociological integration than others, that might change the tone of this paper when it comes to patients seeking companionship.

3.2. Physical

Having BTM will change the way a body develops due to different underlying mechanisms. The deposition of iron into the pituitary gland and the gonads lead to issues during puberty and later infertility. Due to these depositions, patients tend to develop hypogonadism (Lombardo et. al., 2000; Kyriakou and Skordis, 2009; Al-Rimawi et. al., 2005). Due to the tone of this literature review, I believe that some of the internal and external physiological differences a patient with BTM has in regards to others their age was discussed to the depth needed for this paper. However, there were not as many papers as I thought that would deal with other physical manifestation of the effects of the BTM and the effects of the treatment. There was a lot of discussion about shorter stature and infertility, but other physical manifestations were not present in publications.

3.3. Sociology

Although there were numerous papers about the developmental problems that patients with BTM go through, there is a lack of papers on patients in their reproductive years discussing their struggles finding a partner. Patient S discussed how he did not want to disclose his disease status with girlfriends because they would reject him when they found out. However, Patient S was able to get married. Patient S said that this is not the way it usually works out for patients with BTM since many do not get married or have the desire to get married due to their disease status. Part of Patient S's statement was confirmed because one study found that in their sample, only six percent of the adults had been married due to the complications that came with the disease such as cardiological disorders, diabetes mellitus, hepatitis and infertility (Haghpanah et. al., 2013). Although Patient S had a positive outlook on marriage amidst his divorce, a study online discussed that patients with BTM tended to get married if this was viewed as correct in their culture (Wong et. al., 2011). However, this was the only study that approached couples before they were married.

4. Conclusion

This literature review discussed how the differences in psychological and physical development can lead to altered sociological development of a BTM patient with a focus on romantic relationships. After reviewing the literature, many factors contribute to the seemed sociological dysfunction that beta-thalassemia patients exhibit. Patient S's experience was similar in certain aspects, but also different from what was found in the literature in others. However, he mentioned several times in the interview that his experience was unique and not everyone was as lucky. Further research can be done in this field to explore the sociological functioning of BTM patients in regards to seeking out mates which could bring more knowledge to this subject and provide awareness to communities.

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References

- Al-Rimawi, H., Jallad, M., Amarin, Z. and Obeidat, B. (2005) 'Hypothalamic-pituitary-gonadal function in adolescent females with beta-thalassemia major', *International Journal of Gynecology & Obstetrics*, (90)1, pp. 44-47.
- Ansari, S., Baghersalimi, A., Azarkeivan, A., Nojomi, M. and Rad Hassabzadah, A. (2014) 'Quality of life in patients with thalassemia major', *Iran Journal of Pediatric Hematology and Oncology*, 4(2), pp. 57-63.
- De Sanctis, V., Soliman, A. and Yassin, M. (2012) 'An overview of male reproduction in thalassemia', *Rivista Italiana di Medicina dell'Adolescenza*, 10(2), pp. 63-71.
- Galanello, R. and Origa, R. (2010) 'Beta-Thalassemia', *Orphanet Journal of Rare Diseases*, 5(11).
- Ghone, R., Kumbar, K., Suryakar, A., Katkam, R. and Joshi, N. (2008). 'Oxidative stress a disturbance in antioxidant balance in beta thalassemia major', *Indian Journal of Clinical Biochemistry*, 23, pp. 337-340.
- Haghpahanah, S., Nasirabadi, S., Ghaffarpasand, F., Karami, R., Mahmoodi, M., Parand, S. and Karimi, M. (2013) 'Quality of life among Iranian patients with beta-thalassemia major using the SF-36 questionnaire', *Sao Paulo Medical Journal*, 131(3).
- Hakeem, G., Mousa, S., Moustafa, A., Mahgoob, M. and Hassan, E. (2018) 'Health-related quality of life in pediatric and adolescent patients with transfusion-dependent β -thalassemia in upper Egypt (single center study)', *Health and Quality of Life Outcomes*, 16(59).
- Hashemizadeh, H. and Noori, R. (2013) 'Premarital Screening of Beta Thalassemia Minor in north-east of Iran', *Iran Journal of Pediatric Hematology and Oncology*, 3(1), pp 210-215.
- Koutelekos, J. and Haliasos, N. (2013) 'Depression and Thalassemia in children, adolescents, and adults', *Health Science Journal*, 7(3), pp. 239-246.
- Kyriakou, A. and Skordis, N. (2009) 'Thalassemia and Aberrations of Growth and Puberty', *Mediterranean Journal of Infectious Diseases*, 1(1).
- Lombardo, T., Giammusso, B., Frontini, V., D'Arpa, S., Pafumi, C. and Caruso, S. (2000) 'Thalassemic men affected by erectile dysfunction treated with transurethral alprostadil: Case report', *Human Reproduction*, 15(11), pp. 2375-2378.
- Messina, G., Colombo, E., Cassineio, E., Ferri, F., Curti, R., Altamura, C. and Cappellini, M. 'Psychosocial aspects and psychiatric disorders in young adult with thalassemia major', *Internal and Emergency Medicine*, 3(339).
- Miri-Aliabad, G., Fadaee, M., Khajeh, A. and Naderi, M. (2015) 'Marital Status and Fertility in Adult Iranian Patients with β -Thalassemia Major', *Indian Journal of Hematology Blood Transfusions*, 32(1), pp. 110-113.
- Moayeri, H. and Oloomi, Z. (2006) 'Prevalence of Growth and Puberty Failure with Respect to Growth Hormones and Gonadotrophins Secretion in Beta-Thalassemia Major', *Archives of Iranian Medicine*, 9(4), pp. 329-334.
- Naderi, M., reza Hormozi, M., Shrafi, M. and Emamdadi, A. (2012) 'Evaluation of Mental Health and Related Factors among Patients with Beta-thalassemia Major in South East of Iran', *Iran Journal of Psychiatry*, (7)1, pp. 41-51.
- Raiola, G., Galati, M., De Sanctis V., Caruso Nicoletti, M., Pintor C., De Simone, M., Arcuri, V. and Anastasi, S. (2003) 'Growth and puberty in thalassemia major', *Journal of Pediatric Endocrinology and Metabolism*, 16(2), pp. 259-266.
- Rich, A. (1952) 'Studies on the Hemoglobin of Cooley's Anemia and Cooley's Trait', *Proceedings of the National Academy of Sciences of the United States of America*, 38(3), pp. 187-196.
- Roussou, P., Tsagarakis, N., Kountouras, D., Livadas, S. and Diamanti-Kandarakis, E. (2013) 'Beta-Thalassemia Major and Female Fertility: The Role of Iron and Iron-Induced Oxidative Stress', *Anemia*.
- Waheed, F., Fischer, C., Awofeso, A. and Stanley, D. (2016) 'Carrier screening for beta-thalassemia in the Maldives: perceptions of parents of affected children who did not take part in screening and its consequences', *Journal of Community Genetics*, 7(3), pp. 243-253.
- Williams, T. and Weatherall, D. (2012) 'World Distribution, Population Genetics, and Health Burden of the Hemoglobinopathies', *Cold Spring Harbor Perspective Medicine*, 2(9).
- Wong, L., George, E. and Tan, J. (2011) 'Public perceptions and attitudes towards thalassemia: Influencing factors in a multi-racial population', *BMC Public Health*, 11, pp. 193.
- Zeinalian, M., Nobari, R., Moafi, A., Salehi, M. and Hashemizadeh-Chaleshtori, M. (2013) 'Two decades of pre-marital screening for beta-thalassemia in central Iran', *Journal of Community Genetics*, 4(4), pp. 517-522.