Quality of Life Intervention for Individuals with Congenital Abnormality

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Abstract

Children with visible congenital abnormalities often experience negative attention due to the stigma associated with their appearance, which lowers their self-esteem. Currently, psychological treatment for these individuals is not standard practice. Much of the previous research has failed to use a standardized measurement to evaluate the effectiveness of a psychological intervention. This study will evaluate the effectiveness of a digital CBT intervention in helping reduce self-stigmatization in children. It is predicted that children who participate in the therapy will have decreased scores on the Perceived Stigmatization Questionnaire. Approximately 50 affected children between the ages of 8-18 will be recruited from a California hospital using a convenience sample to participate in this study. A between-subjects experimental design will be used, with one group randomly assigned to receive the digital CBT intervention while the other group serves as the control. The participant's scores on the Perceived Stigmatization Questionnaire will be measured after 12 weeks and a t-test will be used to determine if the results are statistically significant. The strengths of this study include the use of a control group to rule out alternative explanations for differences in scores and the inclusion of a broad sample of congenital conditions to improve generalizability. The limitations of this study include limited external validity from being conducted in only one hospital and the use of a self-report measure that limits the reliability of the results.

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Across the world, 8.4 million babies are born each year with a birth defect. (Centers for Disease Control and Prevention, 2023). The high prevalence of birth defects worldwide not only contributes substantially to infant mortality rates but also often creates psychological challenges for those affected. At least 30-40% (Weiss et al., 2006) of these individuals exhibit symptoms that require psychological treatment, however currently only 20% (Hickey & Salter, 2006) of providers perform emotional assessments when evaluating treatment for individuals with facial defects. The purpose of this study is to determine the effectiveness of a digital CBT intervention on the quality of life of patients with visible birth defects. To do this, the different types of abnormality must first be categorized.

Congenital abnormalities are a substantial public health issue that affects millions of humans worldwide. Studies estimate that 3% of babies born in the United States have a birth defect and 6% of babies born worldwide have a congenital anomaly (CA) (Mai et al., 2019). They are a significant contributor to infant mortality, with 94% of related deaths occurring in low and middle-income countries (World Health Organization, 2023). The two main categories of CA are structural and functional, both of which develop during pregnancy and can lead to complications (Abdou et al., 2019). Structural abnormalities are defects in the physical structure of a body part, such as cleft palate, club foot, and missing or abnormal limbs. Many of these are incurable conditions with physical manifestations. Functional abnormalities are defects in the body system itself, including sensory, metabolic, immune, and brain disorders (Birth Defect Research for Children, 2021). They are classified according to severity: minor, severe, or lethal (Czeizel, 2005), with severe and lethal both being considered major abnormalities. Minor

abnormalities affect non-vital organs and do not require immediate correction (Shamim et al., 2010).

Approximately 50% (World Health Organization, 2023) of congenital malformations have no known direct cause, however some known causes are defects within a single gene or chromosome, advanced maternal age, or certain diseases such as the Zika virus or syphilis. Klippel Trenaunay Syndrome, a structural condition that causes port-wine stains, vein malformations, and overgrowth of soft tissue and bones, has been diagnosed as a result of mutations in the AGGF1 and PIK3CA genes (Harnarayan & Harnanan, 2022). These conditions can result in substantial psychological distress. Children born as a result of these diseases exhibit noticeable physical differences that can affect their psychosocial relationships. Understanding this allows society to develop effective treatments to help reduce stigma and discrimination.

Researchers have concluded that stigma is a critical influence on the socioemotional state of individuals born with visible defects (Masnari et al., 2013). Stigma occurs when an individual is perceived negatively, causing a harmful reaction that impacts the quality of their life (Susman, 1994). Stigmatization is typically witnessed in socially discrediting situations of unique individuals based on their identity, such as their ethnicity or sexuality (Weiss et al., 2006). However, health-related stigma is based on an individual's specific health issues, and it aggravates the hidden emotional burden of chronic illness. Frequent manifestations include negative perceptions of self and problems with emotional and social functioning (Masnari et al., 2013), as well as impaired mental health and confidence.

Multiple studies have shown that attractiveness is an important influence on development and interaction (Langlois et al., 2000). Children and adults who are objectively viewed as attractive are treated more positively than those who are considered unattractive. When

compared to children with no physical differences and children with invisible physical differences, children with visible physical differences reported a significantly lower quality of life (Topolski et al., 2005). These individuals experience a range of stigmatizing behaviors from their peers, from staring and avoidance to targeted teasing and bullying (Lawrence et al., 2011). In a society where beauty is regarded as highly important, children and adults with physical disabilities can be viewed as having flaws that diminish their other characteristics, such as intelligence (Green, 2003). The disability wage gap is a prominent example of this aspect of stigma. The most recent studies estimate that North Americans with disabilities make 66 cents for every dollar earned by their non-disabled colleagues (Day & Taylor, 2021). While it is difficult to collect data on recruitment discrimination towards individuals with physical disfigurements, evidence for this statistic comes from studies that show employers prefer attractive candidates over unattractive candidates (Stone & Wright, 2013). The prevalence of this phenomenon highlights a need for an intervention-based approach that can improve patient quality of life.

Interventions aimed at addressing the unique psychological needs of children with congenital abnormalities can improve their quality of life (Hearst, 2007). Given the nonlinear relationship between the severity of the malformation and the amount of self-perceived stigma (Bradbury, 2012), patient needs cannot be generalized. Thus, all healthcare professionals working with these individuals should have a working knowledge of the potential psychological impact. In many cases, only 20% of providers perform emotional assessments when evaluating treatment for individuals with facial defects (Hickey & Salter, 2006). Meanwhile, it is estimated that 30-40% of these individuals exhibit emotional difficulties, such as depression, isolation, and

social anxiety (Weiss et al., 2006). This discrepancy highlights the need for increased awareness and targeted interventions.

Clinical psychologists have proved to be essential in improving the quality of life of individuals with congenital abnormalities (Sarimski, 1998). They can provide emotional support, behavioral interventions, and other cognitive exercise solutions to help children and adults cope with their disabilities. Several current studies propose using clinical psychology methods such as cognitive behavioral therapy (CBT) to help patients improve their quality of life. This is accomplished by assisting patients in reframing their negative thoughts and behaviors and turning them into more positive self-images (Van Dalen et al., 2021). One digital psychosocial intervention called Face IT uses CBT to reduce disfigurement-related distress, improve self-esteem, and increase positive social functioning in individuals with physical differences (Bessell et al., 2012). These outcomes continued to persist at a 3-month and 6-month follow-up, demonstrating the magnitude of their potential impact. Given the remote accessibility, this treatment can be applied on a broad scale and is much more cost-effective than traditional therapies.

In addition to interventions for children, there are also studies showing the importance of interventions supporting their parents and families (Adeyemo et al., 2016). Research estimates between 30-80% of parents with children who have congenital abnormalities experience significant mental health problems, such as emotional distress, anxiety, depression, and isolation (Kolaitis et al., 2017). Interventions that address these issues in parents can help them improve their quality of life, and their children by extension. Studies have found that both parents and children with congenital abnormalities reported improved life satisfaction and decreased stress after participating in a family-centered intervention (Papari et al., 2015). Addressing the unique

mental health needs of both children and their parents can positively influence the quality of life of families affected by congenital abnormalities.

Congenital abnormalities are a significant burden for many patients because they experience stigmatization from their peers that affects their quality of life. CBT interventions have proven effective in improving this measure, as well as anxiety and depression. Previous researchers have not used a standardized scale to measure their findings, nor have they included findings from non-English sources. They also have not been integrated alongside physical treatment for the abnormalities. This study will evaluate responses using a standardized measure. It will evaluate the effectiveness of the treatment when implemented both during physical therapy sessions and when implemented on their own. A digital CBT intervention to reduce self-stigma in patients with visible birth defects will decrease their score on the Perceived Stigmatization Questionnaire compared to patients who do not receive the intervention.

Methods

Participants

The number of participants in this study is anticipated to be 50 children. These individuals will be patients from a large children's hospital in California, United States. Participants will be between the ages of 8-18 with a visible defect and access to a computer with internet, based on inclusion criteria. Exclusion criteria include adolescents who have been diagnosed with depression, body dysmorphia, or a mental or learning disability, as well as children who currently receive face-to-face psychological therapy. The Hispanic/Latino population of California is 40.2%, the Caucasian population is 35.2%, the Asian population is 15.3%, and the African American population is 5.6%, with the remaining 3.7% of the population being American Indian, Native Hawaiian, Multiracial, or Other (USAFacts, 2022). It is

anticipated that the patients will be representative of these percentages. Participants will be recruited using a convenience sample from the patients at the hospital. Each participant will receive monetary compensation of $50 per online session, with each session taking no more than an hour.

Perceived Stigmatization Questionnaire

This measure was developed to measure the impact of stigmatization behaviors that people with a visible defect often experience. It is divided into three subscales: absence of friendly behavior, confused/staring behavior, and hostile behavior (Lawrence et al., 2006). These scales combine into a 21-item inventory with statements such as "People treat me with respect", "People avoid looking at me", and "People call me names". Participant responses are recorded using a 5-point Likert scale (frequencies: 1 = never, 5 = always), with a high score meaning that the individual has a high perception of stigmatization. This measure has high internal reliability with a reported alpha of 0.81.

Demographics

The demographic sheet will ask participants to state their age, gender, ethnicity, year in school, and which congenital abnormality they have been diagnosed with.

Design

This study will be conducted via a between-subjects simple experimental design. The independent variable will be the digital CBT intervention, which 50% of participants will receive, with the other 50% receiving no psychological intervention. The dependent variable will be the participants' scores on the Perceived Stigmatization Questionnaire. All participants will be assigned via randomization to each condition after completing the demographics questionnaire.

Procedure

Participants will be recruited through advertising via the children's hospital with online newsletters will be sent out calling for volunteers. Patients that are interested in participating can then respond and sign up online. Participants will also be recruited via doctors at the hospital, who can notify their patients about the study. Once potential participants sign up on the website, they will be informed of the purpose of the study and given an information packet. They will be notified that participation is voluntary and they can withdraw anytime. They will be given a consent form, with patients younger than 16 years being required to have a parent or guardian sign an additional form. Once the consent form is returned, participants will be given the demographics sheet and the Perceived Stigmatization Questionnaire in order to gather baseline data. They will then be randomized into one of two groups. Group A will complete a CBT intervention session once a week. This will be accessed via the website and participants will work through each session independently. Each session will be monitored weekly to make sure they are completed. Group B will receive weekly care as usual (CAU), wherein they will receive physical therapy care as needed for their congenital abnormality, with no psychological intervention. Participants will be informed that if they are assigned to this group, they will be able to sign up for digital CBT once the experiment is completed, to make this study ethical in that no one ends up being denied treatment. After 12 weeks, both groups will take the Perceived Stigmatization Questionnaire again and their results will be compared.

Data Analysis

In order to determine if a digital CBT intervention reduces self-stigma in patients with visible birth defects, a t-test will be performed. This is appropriate because the dependent variable is continuous, the independent variable is categorical, there is only one IV, and there

are 2 levels in this experiment. If the p-values are at or below 0.05, the results will be considered significant.

Discussion

The purpose of this experiment is to determine if a digital form of cognitive behavioral therapy will have an influence on the self-stigma of children with congenital abnormalities. It is predicted that participants who attend each session of the therapy will score lower on the Perceived Stigmatization Questionnaire than the control group. Comparing the outcomes of children who receive the intervention to those who do not helps rule out alternative explanations for differences in scores between the two groups, which strengthens the validity and reliability of the results. The inclusion of children with a wide range of congenital abnormalities and ages is another strength. Including a broad sample improves generalizability because it allows the results to be applied to a bigger population. However, the overall external validity of the experiment is limited due to being conducted in one hospital in a single geographic area. This means that the results may not be applicable to other cultures or socioeconomic classes. Going forward, researchers could conduct a study across multiple states or countries to increase the sample size and diversity of the study population. The second limitation of the experiment is the use of a self-report measure, which creates the potential for self-report bias and thus limits the reliability of the results. None of the participants are blinded to the purpose of this study, which may lead them to fabricate their answers. However, this measure is necessary for this study to stay within ethical guidelines since it would be dishonorable to use deception for a therapy intervention. In order to improve this in the future, researchers might use more objective physiological or behavioral measures to assess the results of the intervention. Overall, this study provides helpful

insights into the potential effectiveness of a digital cognitive behavioral therapy in reducing the self-stigma of children with congenital abnormalities.

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