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SURGICAL INTERVENTION AND MANAGEMENT OF NURSING CARE IN PAEDIATRIC PATIENTS WITH DIGEORGE SYNDROME

DIGEORGE SENDROMLU ÇOCUK HASTALARDA CERRAHİ MÜDAHALE VE HEMŞİRELİK BAKIM YÖNETİMİ

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ABSTRACT

DiGeorge Syndrome (22q11.2 deletion syndrome) is a genetic disorder affecting multiple organ systems including cardiovascular anomalies, immune deficiencies and developmental delays. Surgical interventions, especially cardiac surgeries, are frequently required in paediatric patients with this syndrome and the importance of a multidisciplinary approach in these processes emerges. In this article, the management of surgical processes in children with DiGeorge Syndrome and the role of nursing care in these processes were examined. The article focuses on critical care areas such as nutrition, immune prophylaxis and infection risk management, as well as the roles of nurses before and after surgery. In addition, the monitoring of complications that may develop due to the characteristics of the syndrome and proactive nursing interventions for these complications are detailed. It is emphasised that nurses, as part of the multidisciplinary team, have a critical role to improve the quality of life of patients and their families, prevent complications and optimise the treatment process. In conclusion, adopting a multidisciplinary approach in the surgical interventions and care processes of paediatric patients with DiGeorge Syndrome is vital in improving patient outcomes and reducing the risk of complications.

Keywords: Paediatric Surgery, DiGeorge Syndrome, Nursing Management.

ÖZET

DiGeorge Sendromu (22q11.2 delesyon sendromu), kardiyovasküler anomaliler, immün yetmezlikler ve gelişimsel gecikmeler gibi çoklu organ sistemlerini etkileyen genetik bir bozukluktur. Bu sendromlu çocuk hastalarda cerrahi müdahaleler, özellikle kardiyak cerrahiler, sıklıkla gerekmekte ve bu süreçlerde multidisipliner bir yaklaşımın önemi ortaya çıkmaktadır. Bu makalede, DiGeorge Sendromlu çocuklarda cerrahi süreçlerin yönetimi ve bu süreçlerde hemşirelik bakımının rolü incelenmiştir. Makale, cerrahi öncesi ve sonrasında hemşirelerin üstlendiği rollerin yanı sıra, beslenme, immün proflaksi ve enfeksiyon riskinin yönetimi gibi kritik bakım alanlarına odaklanmaktadır. Ayrıca, sendromun karakteristik özellikleri nedeniyle gelişebilecek komplikasyonların izlenmesi ve bu komplikasyonlara yönelik proaktif hemşirelik müdahaleleri detaylandırılmıştır. Multidisipliner ekibin bir parçası olarak hemşirelerin, hastaların ve ailelerinin yaşam kalitesini artırmak, komplikasyonları önlemek ve tedavi sürecini optimize etmek için kritik bir role sahip olduğu vurgulanmaktadır. Sonuç olarak, DiGeorge Sendromlu çocuk hastaların cerrahi müdahaleleri ve bakım süreçlerinde multidisipliner bir yaklaşımın benimsenmesi, hasta sonuçlarını iyileştirmede ve komplikasyon riskini azaltmada hayati öneme sahiptir.

Anahtar Kelimeler: Çocuk Cerrahisi, DiGeorge Sendromu, Hemşirelik Yönetimi.

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INTRODUCTION

DiGeorge Syndrome is a genetic anomaly caused by deletion or translocation of a large region on chromosome 22 as a result of a recombination error occurring in gamete cells during meiosis (Lackey et al., 2020). It was first reported as a syndromic disease due to thymus absence in 1829, when the function of the thymus was not fully known (Haskoloğlu et al., 2014). In 1965, Angelo Di George described a group of patients without thymus and parathyroid glands. After this clinical study, patients with 22q11.2 deletions with cardiac defect, immunodeficiency and hypocalcaemia findings were defined as 'DiGeorge Syndrome' (Lackey et al., 2020). De la Chapelle obtained the first genetic source for this syndrome by demonstrating chromosome 22 translocation in 4 family members in 1981 (Haskoloğlu et al., 2014).

DiGeorge syndrome is the most common microdeletion syndrome with a prevalence of approximately 1/4000 -1/6000 live births (Lackey et al., 2020). Although many cases are sporadic (de novo), autosomal dominant inheritance is present in approximately 5-20% of patients. This syndrome is caused by a defect in the development of the 3rd and 4th pharyngeal pockets and migration of neural crest cells in the early embryo period. As a result, pathologies are observed in the pharyngeal arch system including aortic arch branches, cardiac outflow tract, thymus, parathyroid, palate, pharynx and some parts of the face (Göktürk et al., 2016). Approximately 90% of patients have a typical deletion region consisting of ~3 million base pairs and this region contains approximately 90 genes (Lackey et al., 2020). However, phenotype-genotype correlation is very poor in these patients (Göktürk et al., 2016).

The most common clinical findings in patients with DiGeorge syndrome are conotruncal heart defects, immunodeficiency, hypocalcaemia, facial dysmorphism, cleft palate and learning difficulties, while skeletal and urinary system findings are less common. The age at diagnosis of patients with DiGeorge syndrome varies considerably in the literature in accordance with the wide clinical spectrum. Patients are diagnosed at early ages especially in cardiology centres due to their severe clinical pictures, whereas they are diagnosed at late ages in neurology/psychiatry clinics (Haskoloğlu et al, 2014). Surgical interventions are often necessary to improve the quality of life of these patients. However, surgical procedures and postoperative care require a multidisciplinary approach. This article addresses the management of surgical interventions and the importance of nursing care in patients with DiGeorge syndrome.

Digeorge Syndrome and Surgical Interventions

Although DGS has a long history, there are differences of opinion regarding the treatment according to the sources in the medical world. These patients should be handled with a multidisciplinary approach, their phenotypes should be recognised and possible problems should be predicted and each patient should be approached differently according to their needs (Göktürk and Reisli, 2016). Patients hospitalised in the clinic with this diagnosis usually have cardiac problems and may need surgical intervention in the early period (Haskoloğlu and İkincioğulları, 2014).

Heart Anomalies

The most important factors affecting mortality in patients with DiGeorge syndrome are conotruncal heart anomalies and severe immunodeficiency. Approximately 75% of patients have conotruncal heart anomalies due to developmental defects of neural crest cells. Common cardiac anomalies in patients with DiGeorge syndrome include ventricular septal defect (VSD), atrial septal defect (ASD), truncus arteriosus and tetralogy of Fallot. These anomalies usually require surgical intervention (Göktürk et al., 2016).

Tetralogy of Fallot (FT) is the most common heart disease, followed by tetralogy of Fallot with pulmonary atresia, ventricular septal defect, aortic arch and truncus arteriosis. Some congenital heart diseases may be fatal in the intrauterine and perinatal period, but these patients may not survive long enough to investigate the presence of deletion (Göktürk et al., 2016). With advances in surgical techniques and perioperative management, most children born with FT can survive into the adult period of their lives. Recently, studies evaluating both short- and long-term follow-up outcomes after FT operations have identified dramatic improvements in survival since the first operation was performed in 1950. In many centres, perioperative mortality rate is less than 2% with early surgical correction in the

first year of life, and life expectancy over 30 years after surgical repair is reported to be 90% (Al Habib et al., 2010; Moons et al., 2010; Chiu et al., 2012; Valente et al., 2014).

Truncus arteriosus is typically caused by a developmental defect of neuro crest cells and has been experimentally proven. It is therefore often associated with DGS. The prevalence of 22q11.2 deletion is 31% in foetuses with truncus arteriosus and 20-41% in infants and older children (Göktürk et al., 2016). Approximately 80% of patients without surgical treatment are lost within the first year. This pathology, in which pulmonary vascular disease develops rapidly, needs to be repaired in the early period. Developments in intensive care units and the use of safer cardiopulmonary bypass (CPB) methods have increased the success of surgeries performed in the neonatal and early childhood period (Karacı et al., 2012).

The frequency of 22q11.2 deletion in patients with ventricular septal defect varies considerably. Genetic investigation should be performed if other anomalies accompany ventricular septal defect and other characteristic complaints and findings of DGS are present in the patient (Göktürk et al., 2016). Thanks to earlier surgical treatment and increased experience in perioperative patient care, mortality has decreased significantly (<1%) and significant postoperative morbidity has become very rare (Scully et al., 2010; Yıldırım and Aliyev, 2020). Currently, the results of surgical treatment of patients diagnosed with VSD aged less than 6 months are well known. Since the prevalence of pulmonary hypertension in the postoperative period is not well known in older patients, the age at surgery determines mortality and morbidity (Yildirim and Aliyev, 2020).

Endocrinological Anomalies

Features of DGS include absence or hypoplastic thymus, cardiac abnormalities, hypocalcaemia and parathyroid hypoplasia. Hypoparathyroidism can cause symptomatic hypocalcaemia; hypocalcaemic tetany occurs within 24 to 48 hours after birth. Prognosis usually depends on the severity of the heart defect. It is reported that patients with DGS should have their endocrine system reviewed and blood calcium levels checked periodically. If signs of hypothyroidism or hyperthyroidism occur, further investigations should be performed and appropriate treatment should be initiated when deemed necessary (Göktürk and Reisli, 2016).

Urogenital Anomalies

Structural anomalies of the urinary system are found in 30% of patients with DiGeorge syndrome (Wu et al., 2002). Absence of kidney, dysplastic kidney, vesicoureteral reflux, hydronephrosis, nephrocal sinosis, voiding dysfunction, undescended testicle, hypospadias, hydrocele, and ovarian cyst are among the pathologies observed. These pathologies are more common compared to the general population. Patients with DiGeorge syndrome should be screened with radiological imaging for possible pathologies. Anorectal malformations have also been reported (Wu et al., 2002; Bassett et al., 2005).

Immunodeficiency

It is seen in 75% of all DGS cases and develops due to thymus hypoplasia or aplasia. Molecular interaction between thymus epithelium and lymphoid epithelial cells is critical for thymus development. Severe thymus atrophy leads to complete absence of thymocytes and combined immunodeficiency by stopping T lymphocyte development (McDonald-McGinn et al., 2001). Successful results are obtained with bone marrow, peripheral blood and thymus transplantation to reconstitute the immune system (Land et al., 2007; McGhee et al., 2009; Gennery, 2012). Hoehner et al. published the results of thymus transplantation in 44 patients with complete DGS and reported that 72% of the patients achieved immunoconstitution and were alive one year after transplantation. After thymus transplantation, naive and regulatory T cell production with normal T cell repertoire, normal mitogen response, and a limited histocompatibility complex (MHC)-mediated antigen-specific immune response occur (Markert et al., 2007).

Facial Symptoms

Facial dysmorphism, one of the most characteristic features of DGS, is observed in approximately 60-100% of patients (Göktürk et al., 2016). If we look at the characteristic features; a large wide head structure, long forehead, excessive distance between the two eyes (hypertelorism), epicanthal folds, short and wide nose with flattening at the root of the nose, short chin and neck (Özer et al., 2019). Late

onset of speech, phonation disorders, communication and behavioural problems, and learning difficulties are among the problems that occur in advanced periods. Phonation problems may be caused by laryngeal webs, velopharyngeal insufficiency or vocal cord paralysis. Surgery can correct these abnormalities but phonation typically remains problematic. The optimal treatment for speech delay is not fully known (Göktürk et al., 2016). Feeding and swallowing problems due to palatal defects are very challenging for parents in early infancy and may disrupt the developmental stages, the healing process and the body's defence mechanisms. Orogastric/nasogastric tube feeding and cleft palate repair are among the treatment methods. Cleft palate repair can improve feeding ability, speech and reduce the incidence of sinopulmonary infection (Göktürk et al., 2016).

Skeletal System Symptoms

Skeletal system anomalies are between 17 and 47% in patients with DGS. Short neck due to cervical spinal region anomalies; scoliosis due to other vertebral anomalies; extremity anomalies such as gap between the 1st and 2nd toes, small hand and foot, short nail, long/pointed finger, patellar dislocation; hypotonia and pectus excavatum are skeletal system findings (Göktürk et al, 2016).

The Importance of Multidisciplinary Approach

Surgical Team

Surgical interventions in patients with DiGeorge syndrome are vital, especially for patients with serious structural problems such as heart and large vessel anomalies. Conotruncal heart defects, which are frequently seen in this syndrome, should be handled meticulously by the surgical team. The difficulties encountered during surgical interventions can be managed more effectively with the co-operation of a multidisciplinary team (Marino et al., 2001).

The multidisciplinary approach requires the surgical team to work together not only with surgeons but also with anaesthesiologists, cardiologists, intensive care nurses and other relevant health professionals. This team acts in a coordinated manner in all processes from the preoperative assessment of the patient to the complications that may be encountered during surgery and postoperative care. Such co-operation can improve patients' surgical outcomes and reduce the risk of complications (DeMarco et al., 2004). For example, one study reported that surgical procedures performed on patients with DiGeorge syndrome significantly improved surgical outcomes and reduced complication rates when managed with a multidisciplinary team approach. This shows the important role of the surgical team in such complex cases (Marino et al., 2001).

Nursing Role

Nurses have a critical support role for patients living with rare diseases and their caregivers. This role includes providing information about the disease, following medical innovations and continuous efforts to improve treatment processes. Nurses, also as care coordinators and health educators, should provide an education programme that is appropriate to the needs of the patient and their family, free from unnecessary details. They should also support the general well-being of patients and their families by facilitating their access to social and educational support. The nurse should consider the individual as a whole (Pelentsov et al., 2016; Walkowiak and Domaradzki, 2020; Özer and Ay, 2023).

- The patient should be monitored for early signs and symptoms of infection.
- The patient should be observed for signs and symptoms of hypocalcaemia.
- Signs and symptoms of psychiatric disorders should be closely monitored.
- Ensure the safe use of drugs.
- Arrange appropriate speech therapies for patients with feeding and language difficulties.
- Early motor development steps should be closely monitored.

Psychosocial Support

Individuals who have to cope with rare diseases such as DiGeorge syndrome most often face problems such as lack of information, diagnostic difficulties, lack of specialised health professionals, high treatment costs, lack of social support and stigmatisation (Özer & Ay, 2023). The low prevalence of diseases such as DiGeorge syndrome causes patients and caregivers to lack information about appropriate support systems. This lack of information can negatively affect the quality of life of both the patient and the caregiver (Molster et al., 2016). However, this quality of life can be improved through

supportive practices such as participation in patient advocacy groups, ongoing or planned research studies, and access to information about innovations in research protocols (Anderson et al., 2013; Groft and Posada de la Paz, 2017).

Preoperative Nursing Management

Patient Assessment

Preoperative patient assessment is a critical component of nursing management in patients with DiGeorge syndrome. This process includes a comprehensive examination of the patient's general health status, current symptoms and potential risk factors. Conditions such as cardiac anomalies, immune system disorders and hypocalcaemia, which are frequently seen in DiGeorge syndrome, are the main factors that should be carefully considered before surgery. Nurses should review the patient's medical history in detail, gathering information about medications used in the management of existing diseases, allergies and previous surgical interventions. In addition, the psychosocial status of the patient should also be assessed; information should be collected about the concerns, expectations and support systems of family members. Such a comprehensive assessment helps the surgical team to create the most appropriate treatment plan for the patient and helps to prevent possible complications (McDonald-McGinn and Sullivan, 2011). For example, one study reported that detailed preoperative assessments in patients with DiGeorge syndrome contributed to a reduction in postoperative complications and improved overall surgical outcomes (Goldmuntz, 2005).

Education and Information

Preoperative education and information is an important part of nursing management in patients with DiGeorge syndrome. It is important that the parent who applies to the hospital because of a condition related to his/her child has information about his/her child's health. It is necessary to inform the parents correctly before surgical intervention and to provide education with a multidisciplinary approach. Especially the psychological support and correct information provided by the surgical nurse to the parents of children who will undergo surgical intervention reduces the fear, anxiety and uneasiness experienced by the parent and child. The training to be given includes early signs and symptoms of infection, symptoms of hypocalcaemia, safe use of drugs, complications of surgical intervention, immune treatment options, genetic counselling, speech therapy for nutritional and language difficulties, warning signs of delay in early motor development steps, benefits of early intervention programmes, signs and symptoms of psychiatric disorders (Lackey et al., 2020). It is very important for nurses to be aware of the anxiety of pediatric patients and their parents, to determine the causes of anxiety, to take protective measures, to prepare a suitable environment for the implementation of the treatment plan, to demonstrate a holistic and family-centred approach to reduce the negative effects of hospitalisation and surgical intervention on the child and parents (Shields et al., 2006; Çalbayram et al., 2016; Andsoy & Alsawı, 2017).

Immune Prophylaxis

Patients with DiGeorge syndrome require special attention to the risk of infection prior to surgical interventions due to immunological deficiencies. These patients may have poor T cell function due to thymus hypoplasia or aplasia, making them more vulnerable to infections. Therefore, preoperative immune prophylaxis plays an important role to minimise the risk of infection. Before surgery, nurses should perform a thorough assessment of the patient's immune status. This includes T cell counts and function, as well as a review of the patient's current history of infection. When necessary, appropriate antibiotic prophylaxis is administered to patients to prevent intraoperative and postoperative infections. In addition, immune therapies such as IVIG (intravenous immunoglobulin) may be administered to support immune functions. For example, one study showed that immune prophylaxis administered before surgery in patients with DiGeorge syndrome contributed to a significant reduction in postoperative infectious complications (McLean-Tooke et al., 2008; Göktürk et al, 2016).

Postoperative Nursing Management

Monitoring Complications

Because patients with DiGeorge syndrome often carry special risks related to the immune system and cardiovascular system, they may be vulnerable to certain complications in the postoperative period.

Monitoring and managing these complications plays a critical role in nursing management. Paediatric patients with DiGeorge syndrome are at high risk of infection due to immune system deficiencies. Few reports have been published describing the perioperative management of patients with DGS undergoing surgical repair in the presence of congenital anomalies such as congenital heart (Yeoh et al., 2014). However, after surgical interventions in these patients, signs of infection should be carefully monitored; findings such as fever, redness, swelling or discharge from the wound site should be recognised early. Nurses should comply with aseptic techniques and provide appropriate antibiotic treatment to reduce the risk of infection. DiGeorge syndrome is frequently associated with cardiac anomalies, which may increase the risk of complications in the postoperative period. It is important for nurses to carefully monitor for heart rhythm disturbances, high blood pressure or other cardiovascular symptoms. Regular ECG and cardiac monitoring play a critical role in this process. Electrolyte imbalances such as hypocalcaemia, which are frequently seen in these patients, should be carefully monitored in the postoperative period and treated when necessary. Continuous control of electrolyte levels is of great importance for the management of these imbalances (Yeoh et al., 2014; Göktürk et al., 2016; Biggs et al., 2023).

Pain Management

Postoperative pain management is of great importance for children. Inadequate pain management may lead to prolonged hospital stay, patient dissatisfaction and increased morbidity and mortality rates. Therefore, effective postoperative pain management in paediatric patients plays a critical role in accelerating the recovery process and preventing complications (Frizzell et al., 2017). Pain assessment poses a significant challenge in children who cannot express their pain due to age, cognitive impairments or existing diseases. There is no standardised method for the reliable assessment of pain in this group of patients. This deficiency brings with it the risk that pain may not be accurately recognised and its assessment may be inadequate. This may lead to ineffective pain management and undesirable outcomes in treatment processes (Gehdoo, 2004; Andersen et al., 2017). Acting under the guidance of approved clinical guidelines in pain management contributes significantly to effective pain management in the postoperative period. These guidelines set the standards of pain management based on available scientific evidence and clinical experience and ensure that the treatment process is carried out in a systematic and individualised manner. Implementation of approved guidelines can improve consistency and effectiveness in pain management, reduce complications and improve patient satisfaction. In this context, the use of guidelines supports the optimisation of pain management and the improvement of patient outcomes (Okyay and Ayoğlu, 2018).

Nutritional Support

Feeding and swallowing problems in patients with DiGeorge syndrome can pose serious challenges for parents in early infancy. These problems can negatively affect the child's developmental stages, the healing process and the body's defence mechanisms. Strategies such as the use of orogastric or nasogastric tubes, percutaneous endoscopic gastrostomy, or surgical correction of underlying pathologies may be required for the management of feeding problems (Göktürk et al., 2016). These approaches are important steps towards improving feeding and swallowing functions and supporting general health status.

RESULT

Rare and chronic diseases, such as DiGeorge syndrome, require the support of a multidisciplinary healthcare team due to their life-threatening characteristics and difficulties in diagnosis and treatment. In the management of such diseases, each member of the healthcare team has a specific role, and the coordinated fulfilment of these roles directly affects the quality of patient care. Nurses play a critical role in the management of children with DiGeorge syndrome. The fact that nurses have sufficient knowledge and awareness of such rare diseases improves the quality of care at all stages of surgical processes and allows early detection of possible complications. This contributes to maintaining the general well-being of the patient and accelerating the healing process. The active involvement of nurses in the multidisciplinary team is vital for meeting the individual needs of patients and managing the treatment process more effectively.

Conflict of Interest
The authors declare that they have no conflict of interest.

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