Journal of Nursing
Revista de Enfermagem
UFPE On Line
ISSN: 1981-8963

CLINICAL CASE REPORT ARTICLE

NURSING CARE TO CHILDREN WITH CORNELIA DE LANGE SYNDROME

ASSISTÊNCIA DE ENFERMAGEM A CRIANÇA COM A SÍNDROME CORNÉLIA DE LANGE

ASISTENCIA DE ENFERMERÍA AL NIÑO CON EL SÍNDROME DE CORNELIA DE LANGE

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ABSTRACT

***Objective*: to describe the systematization of nursing care to a child with Cornelia de Lange Syndrome. **Method:** a descriptive study with a qualitative approach, of the type study of clinical case, in light of the conceptual model of Horta, performed in a pediatric clinic of a teaching hospital of federal public institution in the city of João Pessoa, Brazil. To collect the data we used the instrument of pediatric clinics, conducted through interviews with the mothers' and children's corporal examination, with the signing of the Term of Consent Free and Lightened, after the approval of the research project by the Committee of Ethics of the University Hospital Lauro Wanderley, of the Federal University of Paraíba and by CAAE No. 0052012600000, under Protocol n ° 222/09. *Results*: nursing care implemented focused on the reducing of discomforts, the monitoring for potential complications, directed, primarily, to the psychobiological needs affected. The nursing identified diagnoses were: Ineffective breathing pattern, Altered nutrition lesser than the body requirements, Risk of aspiration, Hyperthermia, Risk of infection, Delay of the growth and development and knowledge deficit. *Final considerations*: It is expected that this study can help to guide the care to be provided by the nursing team across the carrier of Cornelia de Lange Syndrome. *Descriptors*: nursing process; children; syndrome; pediatric nursing.**

RESUMO

***Objetivo*: descrever a sistematização da assistência de enfermagem a uma criança com a Síndrome Cornélia de Lange. *Método*: estudo descritivo, com abordagem qualitativa, do tipo estudo de caso clínico, à luz do modelo conceitual de Horta, realizado na clínica pediátrica de um hospital-escola de instituição pública federal da cidade de João Pessoa-PB, Brasil. Para a coleta de dados, recorreu-se ao instrumento da clínica pediátrica, realizada por meio de entrevista com a genitora e exame físico da criança, com assinatura do Termo de Consentimento Livre e Esclarecido, após a aprovação do projeto de pesquisa pelo Comitê de Ética do Hospital Universitário Lauro Wanderley, da Universidade Federal da Paraíba e mediante CAAE n° 0052012600000, sob Protocolo n°222/09. *Resultados*: os cuidados de enfermagem implementados focaram a redução de desconfortos, a monitorização de complicações potenciais, direcionadas, prioritariamente, às necessidades psicobiológicas afetadas. Os diagnósticos de enfermagem identificados foram: *Padrão respiratório ineficaz, Nutrição desequilibrada menor do que as necessidades corporais, Risco para aspiração, hipertermia, Risco de Infecção, Atraso do crescimento e desenvolvimento e conhecimento deficiente*. *Considerações finais*: espera-se que este estudo possa contribuir para nortear os cuidados a serem prestados pela equipe de enfermagem frente ao portador da Síndrome Cornélia de Lange. *Descritores*: processos de enfermagem; criança; síndrome; enfermagem pediátrica.**

RESUMEN

***Objetivo:* describir la sistematización de la asistencia de enfermería a un niño con el Síndrome de Cornelia de Lange. *Método:* estudio descriptivo con enfoque cualitativo del tipo estudio de caso clínico, basado en el modelo conceptual de Horta, realizado en la clínica pediátrica de un hospital escuela de una institución pública federal en la ciudad de João Pessoa, estado de Paraíba, Brasil. Para la recopilación de datos se recurrió al instrumento de clínica pediátrica, siendo realizada por medio de entrevista con la genitora y examen físico del niño, con la firma de Formulario de Consentimiento Informado posteriormente a la aprobación del proyecto de investigación por el Comité de Ética del Hospital Universitario Lauro Wanderley de la Universidad Federal de Paraíba y mediante el Certificado de Presentación para Apreciación Ética (CAAE) N° 0052012600000, en virtud del Protocolo N° 222/09. *Resultados:* los cuidados de enfermería aplicados enfocaron la reducción de malestar y el seguimiento de complicaciones potenciales, direccionado prioritariamente a las necesidades psicobiológicas afectadas. Los diagnósticos de enfermería identificados fueron: *Patrón respiratorio ineficiente; Nutrición desequilibrada menor que las necesidades corporales; Riesgo para la aspiración; Hipertermia; Riesgo de infección; Atraso de crecimiento y desarrollo; y Conocimiento deficiente*. *Consideraciones finales:* se espera que este estudio pueda contribuir a orientar los cuidados prestados por el equipo de enfermería al portador del Síndrome de Cornelia de Lange. *Descriptores:* procesos de enfermería; niño; síndrome; enfermería pediátrica.**

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INTRODUCTION

The Cornelia de Lange Syndrome (SCL), also called Brachmann de Lange syndrome, is a rare congenital anomaly multiple, with an incidence of 1:10,000 to 1:30,000 in case births and whose clinical picture depends on the intensity, the number of affected segments, the level of dysfunction and aggressiveness of genetic involvement. In most cases, the bearer of this syndrome has low weight, microcephaly, small limbs, clinodactyly, intergital membrane and single palmar crease.

The diagnosis of this syndrome is based on clinical findings. There is no biochemical or chromosomal markers that aid in the diagnosis of the disease, which complicates the determination of its incidence in the population. In general, facial dimorphism displayed by carrier rounded face, eyebrows together, elongated eyelashes, small nose and thin lips and slightly inverted) is sufficient to suspect the diagnosis.

The prognosis is stationary, but common infections in infancy, hypotonia and severe bowel obstruction may complicate the clinical picture. Other potential complications are congenital heart defects, gastroesophageal disorders, joint changes, hormonal, ocular and seizures. Regarding the mortality rate is high and almost three times that of the general population. The most common causes of death are pneumonia, followed by digestive and cardiac defects.

In this context, we highlight the importance of the role of the nurse in assisting the needs of the carrier's syndrome Cornelia de Lange and his family. For this type of care, the nurse should use the nursing process as a work tool. This process is a method of dynamic, flexible and organized, that is, a way by which the nurse collects data, identifies needs professional care, proposes interventions and evaluates patient outcomes for interventions implemented, which favors Care System Nursing SAE).

In everyday practice, incorporating care favors the systematic documentation of the care provided and contributes to communication between members of the nursing team and continuity of care. The documentation based on the nursing process generates data that can describe and explain the phenomena by which nurses are accountable, demonstrating thereby the visibility and contribution of nursing in health care. Moreover, the information generated by the data of Nursing can be used for research, teaching and administration. In this perspective, the SAE is an instrument of assistance and guidance of professional autonomy, resulting from the application of the concepts of a conceptual model, through the operation of the nursing process.

To guide the nursing care was used Horta's Conceptual Model which according to the concepts and assumptions the object of nursing care is the satisfaction of basic human needs individual, family and group. Human needs are generated by states of imbalance in time and space, that drive the individual to seek ways to restore balance organic.

According to this theoretical perspective, the nurse is responsible for the maintenance of equilibrium states, prevention and reversal of imbalances through human assistance in meeting their basic needs, using, for this, knowledge and techniques accumulated in Nursing.

Regarding the SCL, there is a lack of studies in the literature that focus on nursing care directed to the various needs affected by the syndrome. Therefore, we must collaborate to develop studies that nurses knowledge, in order to improve the quality of life of patients and improving the care of a multidisciplinary team.

The objective of the study was to describe the systematization of nursing care to a child with the SCL, the Theory of Basic Human Needs Horta.

METHOD

This is a descriptive, qualitative, structured approach to clinical case study, for which we employed the nursing process as a method. We traversed the stages of data collection, nursing diagnosis, planning, intervention and evaluation of results, in light of the Basic Human Needs.

The case study is suitable to be applied in direct nursing care in order to investigate deeply the problems and needs of the person, family or community, which enables the development of strategies to address or reverse the problems found .

At the stage of History of Nursing, conducted with survey data that would enable the child to identify nursing problems, needs affected. To collect data, we used the instrument built for the pediatric clinic, which favors the systematic record of the interview and physical examination, in categorical basic human needs (psychobiological, psychosocial

and psychospiritual).

Then the data were analyzed, in order to diagnose the problems of nursing that need the intervention of the nurse to restore balance in time and space and the degree of dependence of the child and his mother in relation to nursing staff.

At this stage, followed the following steps: reading the case, identification of significant data; reading of meaningful data; collation of data and identification of nursing diagnoses, using the nomenclature of NANDA diagnoses Internacional.15 From the determined settled the expected results with the assistance and nursing interventions targeted to the needs of affected children to achieve these results. To plan interventions, we took into account the degree of dependency of the child and his mother, in nature and in scope.

The evolution of the child care provided was based on the records made during his hospitalization and daily assessment performed by the researchers who followed and watched.

The study was conducted with the participation of a child with Cornelia de Lange Syndrome, admitted to the Pediatric Clinic of the University Hospital Lauro Wanderley, Federal University of Paraíba / HULW / UFPB. Before the study, we requested permission from his mother to sign the Instrument of Consent. It was explicitly stated that participation in the survey would be voluntary and that she could give up at any time, without prejudice, if so desired, in accordance with Guidelines and Standards for Research Involving Human contemplated in Resolution 196/96.

This study is linked to a master research project, which is being developed in HULW / UFPP, with the approval of the Research Ethics Committee under protocol number 222/09.

RESULTS AND DISCUSSION

♦ History of Nursing

Ablactente twelve months, natural Nazarezinho - PB, coming from the same county, was admitted to the Pediatric Clinic for treatment of congenital heart defects, gastroesophageal reflux disease and conjunctivitis, accompanied by his mother, whose diagnosis the SCL. According to his mother, he was born pre-term, through surgical delivery, low birth weight (1.250g), 39 cm long, with cleft palate, gastroesophageal reflux, poor sucking in breastfeeding and no history of congenital syndromes. Had rounded face, thin eyebrows and united in the midline, elongated eyelashes, small nose, thin lips, microcephaly, small limbs, and clinodactyly membrane intergital the third toe of both feet, absence of teeth and language delay (not speaking) - did not respond to verbal requests to look. Sleep and rest satisfactory; hygiene preserved, but oral hygiene, impaired. Accepted 60% of the diet offered. When the physical examination: general condition regular, pale mucous membranes and skin (+ / 4 +), hydrated with turgor preserved, acyanotic, anicteric, fever (axillary temperature of 38.6 ° C); tachycardic, regular heart rhythm in two days , heart sounds normal with murmurs diffuse peripheral perfusion and tissue preserved taquiesfígmico, blood pressure (BP) = 90 x 60 mmHg; tachydyspnea, audible breath sounds, adventitious sounds, chest expansion bilaterally diminished; plan abdomen, normotensive, painless to superficial and deep palpation, bowel sounds present in all four quadrants; eliminations bowel and bladder present and normal (SIC); motor hypotonia, without coordination. Imaging studies revealed: congenital heart disease with pulmonary valve stenosis, and abdominal ultrasonography total unchanged.

♦ Planning, implementation and evaluation of nursing care

The nursing care provided to children with SCL, in light of the conceptual model of Horta, focuses on identifying the basic needs affected by the disease, because if your needs are not being addressed or inadequately addressed, this creates discomfort, if prolonged, contributes to its sickness. Thus, nursing care planned and implemented were focused on reducing discomfort, monitoring of potential complications, promoting quality of life, within the possibilities and the child's individuality.

The most common problems in patients with this syndrome, generating imbalances in time and space, including language delay, cardiac abnormalities, gastroesophageal reflux, feeding difficulties and growth changes, speech and development psychomotor.

The child in question is a dependent mother, not only because of his medical condition, but also in relation to their age, which makes it dependent on the nursing staff. Accordingly, the planned assistance included both the child and his mother / caregiver.

The nursing diagnoses identified in children

and mothers' were ineffective breathing pattern; imbalanced nutrition less than body requirements; risk of aspiration; hyperthermia; retarded growth and development, risk of infection and poor knowledge of the mother (Figure 1). It is observed that nursing diagnoses are mainly related to physiological needs, altered by genetic syndrome.

Nursing diagnoses	Expected results	Nursing interventions
Ineffective breathing pattern	Must present decreased respiratory effort	1. Position in Fowler; 2. Administer oxygen if necessary; 3. Monitor signs of cyanosis.
Imbalanced nutrition less than body requirements	Must present weight gain during hospitalization.	1. Offer fractional diet (in smaller quantities and higher frequency); 2. Weigh daily, to evaluate weight gain or loss; 3. Investigate food preferences and notify the service of nutrition; 4. Guide the mother about the correct position to feed the child; 5. Provide the milk antireflux. 6. Raise the Food Administration after decubitus, in order to provide better digestion and prevent reflux.
Risk for aspiration	Will be aspirations to the lower respiratory tract.	1. Guide the mother about the need to put the child to eliminate gases after the diet; 2. Raise the decubitus after administration; 3. Place the child in lateral decubitus, to avoid aspiration of secretions. 4. Guide the mother about oral hygiene.
Hyperthermia	Must present temperature decrease to normal parameters	1. Administer medication as prescription; 2. check the temperature of 4/4:0 or when necessary; 3. Promote an airy environment; 4. Guide the mother regarding the use of cold compresses if persistence of fever; 5. Check temperature 30 minutes after administration of antipyretic medication.
Delayed growth and development	Should present better cognitive and motor development, through stimulation of a multidisciplinary team	1. Verify with the medical team clinical follow-up request with speech therapist, occupational therapist and motor therapy; 2.Orient the mother using colorful toys and sounds that draw attention of the child; 3. Enroll in motor and cognitive evolution Handbook.
Risk of infection	Should not present the period of hospitalization associated infections.	1. Monitor signs of infection (fever, chills, cough, shortness of breath, oral pain or swallowing, tachypnea, tachycardia) 2. Monitor the count of leukocytes; 3. Maintain aseptic technique in performing invasive procedures (venous puncture and parenteral medication intramuscular)
Deficient knowledge of the mother on the pathology and child care.	The mother should demonstrate better knowledge about the pathology and child care	1. Inform the mother regarding the pathology, diagnosis and nursing care that must be performed; 2. Ask questions of mother for child care; 3. Guide the mother how to care and special dedication with the child, due to limitations from pathology; 4. Offer psychological support to the mother.

Figure 1. Planning of nursing care to a holder of Cornelia de Lange Syndrome. João Pessoa, 2009.

Nursing diagnosis Ineffective breathing pattern, related to the presence of cleft palate, evidenced by respiratory effort and tachydyspnea moderate, was identified at the time of data collection. To meet this need affected were asked the following interventions: the child was positioned in elevated head (Fowler) and was administered oxygen, resulting in the improvement of dyspnea in the regulation of respiration and the absence of cyanosis. During hospitalization, the child's breathing pattern ranged from mild dyspnea and eupneia. It is believed that this is due to difficulty breathing cleft palate and related muscle hypotonia weakness of the respiratory muscles.

In children in the study, diagnosis Altered nutrition less than body requirements related to the presence of gastroesophageal reflux disease, evidenced by low body weight for his age. Gastroesophageal reflux disease is a complication of high frequency in patients with Cornelia de Lange Syndrome, which can be present in about 77% of cases.The possibility of death from apnea or aspiration determines the need to monitor these phenomena and advice on the care and therapeutic food.

To encourage the child's weight gain in the study, compared to the present framework of malnutrition during hospitalization was

offered antireflux milk, and other food stands in fractional amount every four hours.

The mother was instructed to place the child in decubitus high during the experimental diet and thereafter, to prevent gastroesophageal reflux and decrease the risk of aspiration. Furthermore, was oriented on the importance of eliminating the gases after gastric administration of antireflux milk and maintain the child in the lateral position during sleep.

The child had decreased episodes of gastroesophageal reflux after meals. In nineteen days, there was a weight gain of 500g. During hospitalization, the child showed no respiratory infections secondary to aspiration.

Before being hospitalized, the child had been experiencing recurrent episodes of hyperthermia. On admission, the picture hyperthermic led to suspicion of infection, which was dismissed based on the results of laboratory tests. However, the child kept recurring frames hyperthermia during hospitalization. The literature shows that people with this illness can register inefficient thermoregulation associated with neurological abnormalities.

Nursing diagnosis Hyperthermia was identified based on altered metabolic rate, low infant weight and axillary temperature above the reference values. To reduce elevated body temperature, and the administration of antipyretic, were provided to mother cold compresses for positioning in the frontal, axillary and femoral.

Changes in growth, speech and psychomotor development are common in children with Cornelia de Lange syndrome and were found on the child in question. Besides, she did not follow objects with their eyes, nor sought the direction of the sounds, which allowed the identification of diagnostic delay of growth and development, as evidenced by the difficulty of performing motor activities, social and expressive language typical of his age and cognition). To evaluate these activities were used as parameters the data contained in the Handbook of Children, proposed by the Ministry of Health, according to which, around three to four months, the child follows objects with his eyes, searching the direction of sounds and, 12 months, current age of the child in the study, claps, stands, can hold objects with the index finger and thumb, and say a few words.

The cognitive profile of patients with this syndrome include delays in speech, language in some cases minimal or absent. In all patients, the expressive language is more affected than verbal comprehension and response, while the organization of perception and visual and spatial memory is normal. These patients may have attention deficit or decrease sensory and deafness). According to some authors, the carriers of SCL have delayed mental development, however, have a memory and visual-spatial perceptual organization at a higher level than the other facets.

In this sense, the child's mother was advised to use colorful sound toys, in order to draw the child's attention, to stimulate their motor and cognitive development. Additionally, physical therapy helped during this process, through passive exercise to prevent muscular atrophy. Already Speech acted in the best performance muscle of the articulators. As a result, one can observe a slight improvement in the movement of the lower limbs and the child followed, gradually, the objects with their eyes, showing more attention. She appeared more active and cooperative, with considerable evidence of cognitive evolution, for example, reached some objects with her eyes.

Diagnosis Risk of infection was identified from the following risk factors: gastroesophageal reflux disease, malnutrition and invasive procedures. In this sense, the goal of nursing is the absence of infection during hospitalization. Therefore, the actions implemented included the monitoring of signs of infection and laboratory tests, as well as the performance of procedures by nursing staff rigorously aseptic.

The Cornelia de Lange Syndrome is a rare condition and rarely described in the national literature. Ignorance about the syndrome and care required by the child was expected progenitor, since it was also to the researchers before the study. The sufferer does not achieve autonomy over their care and life activities diária17, which makes it imperative that caregivers have sufficient knowledge to meet your needs. Given the above, the diagnosis was identified Deficit knowledge of mother.

To achieve positive results in relation to the mothers' education, the researchers had to find out about the syndrome and its influence on the basic needs of the child, that the mother was properly oriented in relation to the care of their child during hospitalization and in his later period . She received guidance on congenital and hereditary condition of his son, the changes

associated with cognitive-behavioral, its limitations and the care that needed to be implemented to meet your needs. This approach favored its tranquility regarding pathology and science of their dedication, patience and care with your child.

Implemented stocks mentioned, it was observed that the progenitor seized the guidance given and demonstrated interest in adopting behaviors that previously were not part of routine, for example, more careful oral hygiene of children. The oral care are essential to detect and correct dental abnormalities, since the reflux of stomach acid causes caries, gingivitis, micro growth and erosion dentes.17 Normally, the teeth begins at six months of age, but children under study had no teeth, despite having twelve months of age.

FINAL THOUGHTS

Caring for a child with a rare syndrome has been challenging to the researchers, because the approach is not about the disease during graduation, but a lack of studies on the topic, especially with regard to nursing care.

The operationalization of concepts and theoretical assumptions of Horta, using the nursing process, allowed the targeting of nursing care for the basic needs of the affected child with the SCL. The use of the nursing process favored a systematic care, both in actions and in documentation, allowing researchers to apply a scientific method to solve problems presented in the light of a theoretical framework.

In this study, the results showed that assist a child with the SCL, directed to prevent potential complications arising from their clinical condition and their cognitive and behavioral aspects, contributed to an improvement in the condition of the patient, which presented more cooperative, peaceful and showed a slight improvement in their nutritional status.

It is believed that an important fact to achieve the results was the empathic relationship built between the caregiver (nurse) and the cared for (child) because there was a relationship of mutual respect and trust. It was felt that to assist qualified nursing and humanized, nurses need to use beyond the standard of empirical knowledge, personal standards, ethical and aesthetic, the art of nursing.

Finally, it is hoped that this study can help guide the care to be provided by the nursing team ahead bearer of Cornelia de Lange Syndrome, since the rarity of the disease, associated with little knowledge leads to an assistance nursing intuitive, disjointed theoretical knowledge.

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Sources of funding: No
Conflict of interest: No
Date of first submission: 2012/03/30
Last received: 2012/21/21
Accepted: 2012/09/22
Publishing: 2012/10/01

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