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Sturge-Weber Syndrome: The importance of comprehensive care in assisting adolescent patients

Síndrome de Sturge-Weber: la importancia de la atención integral en la asistencia a pacientes adolescentes

Síndrome de Sturge-Weber: A importância do cuidado integral na assistência ao adolescente portador

ABSTRACT

Objectives: To describe the experience of nursing students during the care of adolescents with Sturge-Weber Syndrome and the importance of comprehensive care. **Method:** This is a descriptive, exploratory research, with a qualitative approach, of the type of experience report, carried out in a School Clinic in the Mountain Region of the State of Rio de Janeiro, in the month of June 2019. **Result:** BHV, 13 years old, male, attending 4th year of elementary school, with Sturge Weber Syndrome, mentions that he had not been to a health service and routine exams for a long time. It had a "spot on the face", characteristic of the syndrome. He has glaucoma and uses Phenobarbital due to the seizure. He is afraid because of "the stain on his face". The data indicate the need for comprehensive care. **Discourse:** As it is a rare disease, the need for comprehensive care is evident, due to neurological, cutaneous, ocular and oral manifestations. Our conduct allowed us to return to ophthalmic care, dental follow-up, clinical and neurological evaluation. **Final considerations:** It was possible to realize that in order to achieve quality in comprehensive care in the care of adolescents with a rare syndrome, such as that of Sturge-Weber, the involvement of a multidisciplinary team is extremely important.

DESCRIPTORS: Sturge-Weber Syndrome; Nursing Consultation; Adolescent; Multiprofessional Team.

RESUMEN

Objetivos: Describir la experiencia de los estudiantes de enfermería durante el cuidado de adolescentes con Síndrome de Sturge-Weber y la importancia de la atención integral. **Método:** Esta es una investigación descriptiva, exploratoria, con un enfoque cualitativo, del tipo de informe de experiencia, realizado en una Clínica Escolar en la Región Montañosa del Estado de Río de Janeiro, en el mes de junio de 2019. **Resultado:** BHV, 13 años, hombre, que cursa el cuarto año de la escuela primaria, con el Síndrome de Sturge Weber, menciona que no había estado en un servicio de salud y exámenes de rutina durante mucho tiempo. Tenía una "mancha en la cara", característica del Síndrome. Tiene glaucoma y usa fenobarbital debido a la convulsión. Tiene miedo debido a "la mancha en su rostro". Los datos indican la necesidad de una atención integral. **Discurso:** como es una enfermedad rara, la necesidad de atención integral es evidente debido a las manifestaciones neurológicas, cutáneas, oculares y orales. Nuestra conducta nos ha permitido volver al cuidado de los ojos, el seguimiento dental, la evaluación clínica y neurológica. **Consideraciones finales:** fue posible darse cuenta de que para lograr la calidad en la atención integral en la atención de adolescentes con un Síndrome raro, como el de Sturge-Weber, la participación de un equipo multidisciplinario es extremadamente importante.

DESCRIPTORES: Síndrome de Sturge-Weber; Consulta de Enfermería; Adolescente Equipo Multiprofesional.

RESUMO

Objetivos: Descrever a experiência vivenciada por acadêmicos de enfermagem durante o atendimento ao adolescente com Síndrome de Sturge-Weber e a importância do cuidado integral. **Método:** Trata-se de uma pesquisa descritiva, exploratória, com abordagem qualitativa, do tipo relato de experiência, realizada em um Ambulatório Escola na Região Serrana do Estado do Rio de Janeiro, no mês de junho de 2019. **Resultado:** BHV, 13 anos, sexo masculino, cursando 4º ano do Ensino Fundamental, portador da Síndrome de Sturge Weber, menciona que há muito tempo não realizava consulta em um serviço de saúde e exames de rotina. Apresentava, "mancha na face", característica da Síndrome. Possui glaucoma e utiliza Fenobarbital devido à crise convulsiva. Apresenta receio devido "a mancha na face". Os dados sinalizam a necessidade de um cuidado integral. **Discussão:** Por ser uma doença rara, fica notório a necessidade do cuidado integral, devido as manifestações neurológicas, cutâneas, oculares e orais. Nossa conduta proporcionou retornar aos cuidados oftalmológicos, acompanhamento odontológico, avaliação clínica e neurológica. **Considerações finais:** Foi possível perceber que para atingir a qualidade no cuidado integral na assistência ao adolescente com uma Síndrome rara, como a de Sturge-Weber é de extrema importância o envolvimento de uma equipe multidisciplinar.

DESCRIPTORIOS: Síndrome de Sturge-Weber; Consulta de Enfermagem; Adolescente; Equipe Multiprofissional.

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Simone Fátima de Azevedo

Teaching nurse at Colégio Santa Catarina - Petrópolis. Preceptor of the Adolescent Nursing Module at the School Outpatient Clinic, by the Faculty Arthur Sá Earp Neto (FASE). Postgraduate in Nursing in Pediatrics and Neonatology. <https://orcid.org/0000-0003-0581-7063>

Lívia da Silva Firmino dos Santos

Coordinator of the Supervised Internship in the Outpatient Network of the Nursing Course at Faculdade Arthur Sá Earp Neto - FASE at the Escola FASE Outpatient Clinic. Master's in health care sciences Postgraduate in Occupational Nursing. <https://orcid.org/0000-0002-9740-1956>

Rafael Ribeiro Rampini

Graduating in Nursing from the Teaching Institution Faculdade Arthur Sá Earp Neto FASE. <https://orcid.org/0000-0003-4779-3645>

Ana Carolina Ferreira da Silva

Graduating in Nursing from the Teaching Institution Faculdade Arthur Sá Earp Neto FASE. <https://orcid.org/0000-0001-7231-0882>

Mariana Ferreira dos Santos

Graduating in Nursing from the Teaching Institution Faculdade Arthur Sá Earp Neto FASE. <https://orcid.org/0000-0002-3764-3865>

Márcio José Barbosa Bertholy

Nurse. Graduated in Nursing from the Teaching Institution Faculdade Arthur Sá Earp Neto FASE. <https://orcid.org/0000-0003-0620-4832>

INTRODUCTION

Sturge-Weber Syndrome (SSW) originates from a genetic alteration, around the sixth week of gestation, where the mutation of the GNAQ gene responsible for the normal vascular maturation process occurs, this mutation allows for a failure in the regression of the primitive cephalic venous plexus, interfering with the normal development of the face, eyes and brain, generating the main signs and symptoms that characterize the disease^(1,2).

Therefore, it is characterized by generating vascular malformations at the cerebral, facial and ocular level, Sturge-Weber Syndrome is considered a rare disease with a worldwide incidence of one case in 50,000 individuals^(3,4).

A typical characteristic of this disease is the port wine stain, which usually appears in the first years of life, a purple/reddish stain with well-defined margins. The spot tends to be in areas innervated by the trigeminal nerve, extending from the ophthalmic branch to the mandible, unilaterally or bilaterally. Over the years,

the stain may increase in thickness and intensity of staining, even generating possible angiomatous nodules^(1,5).

The clinical picture of the disease includes, in addition to the port wine stain, ocular and central nervous system involvement, generating low visual acuity, hemianopsia, scleral, conjunctival or choroidal hemangioma, glaucoma, and leptomeningeal hemangioma that can cause hypertrophy of the cerebral and parenchyma cortical and subcortical calcification causing seizures, developmental delay, intellectual disability, learning disability, hemiparesis and headache^(1,2).

The syndrome can also cause changes in the oral cavity through angiomatoses, causing gingival hyperplasia, macroglossia, and other orthodontic changes that increase the risk of bleeding during dental procedures or intubations^(5,6).

Sturge-Weber Syndrome can be classified into three types, according to the Roach scale: Type I, characterized by the presence of the port wine stain and leptomeningeal hemangioma, with or without glaucoma; Type II, with the presence of the port wine stain and pre-existing

glaucoma; And Type III, with leptomeningeal hemangioma with or without the presence of glaucoma⁽²⁾.

In the case of a disease that involves multiple variables, comprehensive care for the patient with the syndrome is of fundamental importance. Comprehensive care permeates the condition that it is exclusively a principle of SUS, its dimension is seen as a value expressed by health professionals in their care practice through the interaction between the professional and the patient⁽⁷⁾.

One of the essential actors in the implementation of the principle of integrality in their professional practice is the nursing professionals, who in the assistance provided to the healthy or sick individual contribute to the evaluation and proposals of health care strategies for children and adolescents at different levels of health assistance⁽⁸⁾.

In this sense, it is worth mentioning that adolescence is a period of development that occurs between childhood and adulthood, this phase is marked by bodily, social, emotional and intellectual changes during the 10 to 19 years of age,

having complex characteristics, where numerous phenomena occur, such as the search for an identity, the valorization of social groups and possible departure from family values, religious crises, the development of sexuality, in order to build their own personality^(9,10).

This study aims to answer the following guiding question: How to provide comprehensive care in assisting adolescents with Sturge-Weber syndrome?

According to the above, the objective of the present study is to describe the experience of nursing students during the care of adolescents with Sturge-Weber Syndrome and the importance of comprehensive care.

METHODOLOGY

This is a descriptive, exploratory research, with a qualitative approach, of the experience report type, carried out at the School Ambulatory of a Private Higher Education Institution, in the Mountain Region of the State of Rio de Janeiro, in June 2019, included in the on 12/06/2019.

The rapporteur was based on the experience of students, from the 7th and 9th period, of the Bachelor of Nursing Course, while taking the course "Supervised Internship in an Outpatient Network" in the Adolescent Health Nursing Module, under the supervision of the Preceptor Nurse.

And the experience during the care of the adolescent patient with Sturge-Weber Syndrome, clarifying doubts and acting as a mediator during this process to offer comprehensive care, activating the multidisciplinary team to assist the adolescent with this rare syndrome.

The adolescent's nursing service has a partnership with several educational institutions, and one of them is a Municipal School close to the Health Unit, where, at the request of the principal, activities in health education are carried out and students are directed to consultations at the clinic. The activities carried out by nursing students, from the Adolescent

Nursing Module, are supervised by the Preceptor Nurse, and the students benefited are from the 4th and 5th grades of Elementary School.

It is worth mentioning that, since it is an experience report, there was no need to submit it to the Research Ethics Committee, but the authors respected the ethical aspects in preparing it.

EXPERIENCE REPORT

At the Nursing consultation, BHV, a teenager, 13 years old, male, attending 4th grade of elementary school, attended accompanied by his father, sent by the Municipal School where nursing students carry out educational actions in health.

At first, during the reception, the academic observed that the adolescent presented, "spot on the face". Father reported that his son has Sturge Weber Syndrome and had not been in a health service for a long time and had routine tests.

During the adolescent's nursing consultation, students perform an approach with a global view, consisting of the following steps: anamnesis, physical examination, guidelines and referrals, if necessary. At the time of anamnesis, the first consultation instrument is completed, where the adolescent and the family member present the data collection: identification, personal history, family history, family data, housing conditions, eating habits/water intake and elimination of intestinal vesicles.

Currently, academics were able to identify that the teenager has glaucoma and uses Phenobarbital. They also identified that the teenager was hospitalized shortly after birth due to Sturge-Weber Syndrome and at 03 years old he was hospitalized again, due to the seizure crisis, remaining under hospital care for 23 days. It was during this period that phenobarbital 40mg (twice a day) was prescribed, and after using the medication, there were no new episodes of seizure. She has been using eye drops since she was two years old, currently using Cosopt

(one drop in each eye), due to glaucoma in her left eye.

In the second moment of the anamnesis, still under the supervision of the preceptor, students are alone with the adolescent, as it is the most important moment of the consultation, since it is the opportunity for the adolescent to feel safe to expose confidential issues and for the student reinforce the bond created in the previous step. At that moment, the student asked about: issues of puberty (gynecological-urological) and sexuality, self-image and body acceptance, psycho-emotional processes, school space and education, work, social life, life habits, visual and hearing acuity and vaccine booklet.

What draws the attention of academics, even due to issues related to the syndrome, is that the patient denies difficulty viewing the picture during class and hearing difficulties, however, he reports three failures in school years. She says she likes to study and attend classes regularly. He has severe difficulty in reading and little writing, uses short and simple words. Despite the age group, he is still starting to paint figures within the inner limit of the image and reports feeling more secure and firm when using a pen. He is active, communicative, outgoing, reports good interaction with schoolmates and the street where he lives. He plays football on Saturdays and plays video games. Has outdated vaccination card. He has high self-esteem and although he is excited to start taking the bus to his new school in 2020, he is afraid due to "the stain on his face" and having to explain his pathology to his new schoolmates.

The academics signal concern about the fact that the patient was, until then, in a safe and well-known environment, where there was no problem with the characteristics of the disease that is the stain of port wine in the face, but the new school may bring discomfort due to colleagues' view.

The physical examination showed the following parameters: WEIGHT:

41,200 kg; HEIGHT: 1,51m; BMI: 18.07; TAX: 35.9; FR: 16 rpm; HR: 76 bpm; PA: 120/60 mmHg; Tanner staging: P2 and G1. The data were classified as appropriate for the age, according to the Adolescent Handbook of the Ministry of Health⁽¹¹⁾.

Skin integrity without changes and skin color according to Syndrome features; superficial and deep palpation in the abdomen without changes or palpable masses; hyperpigmentation was observed in the upper and lower quadrants, which extends to the lower limbs where the coloring normalizes.

At the end of the consultation, the academics highlighted the following nursing diagnoses as the greatest risk: 1-Risk of low situational self-esteem related to changes in body image. 2-Impaired dentition characterized by dental caries related to inadequate oral hygiene.

They followed the consultation with the following guidelines, regarding: update of the vaccination booklet; importance of oral hygiene; importance of dedicating oneself to studies; perspective for the future; bullying; about anxiety about the new school and encouraged to remain with high self-esteem; non-involvement with legal and illegal drugs;

To close the consultation and meet the adolescent's demand, according to the nursing diagnosis found, the students and the governess opted for the following conduct: referrals to the following services: ophthalmology; neuropsychiatry; medical clinic; dentistry; vaccine room to administer the first dose of HPV and meningococcal vaccine; requesting laboratory tests of the adolescent's routine and scheduling a return appointment with the nurse.

The preceptor and students understood that to provide comprehensive care in the care of adolescents with Sturge-Weber Syndrome it would be necessary, not only the guidelines on general health care during adolescence, but also the participation of a multidisciplinary team, to thus bringing resolution to the patient's needs and consequently avoiding future

complications. In this way, professionals from each specialty were contacted and after passing the case, the following outcomes were achieved:

- Ophthalmologist - attended on 06/19/2019, a teenager arrived accompanied by "the mother of the heart" (SIC), says a history of glaucoma, mentioned that he was being monitored in Rio, at UFRJ. At the time of the consultation, he complained of difficulty reading from a distance. In daily use of eye drops for 02 years of age. On physical examination, hemangioma in hemiface D (mainly) and E. In ophthalmologic evaluation: AV: OD = NPL and OE = 20/20 ct (+1.00 sp / 1.50 cyl at 170°). Bio: Od: white cataract, posterior synechiae; band keratopathy. OE: no changes; IOP = 30/30, FO: OD: unavoidable, OE: DO esc 0.5 and macula without changes, regular vessels, retina applied without evident retinal lesions. Conduct: New eye drops prescribed in OE, and sent for consultation at the Ophthalmology Service of Pedro Ernesto University Hospital - HUPE (UERJ).
- Neuropsychiatrist - attended on 06/26/2019 at 10:30 am, reports using 40 mg phenobarbital (twice a day); last convulsive crisis at 03 years old, difficult to control; episode of sudden right hemiparesis; last image exam at 03 years old. On physical examination: flat hemangioma of the face with a predominance of D, enlargement of soft tissues, hemiface D and tongue; no focal motor deficit, but little fine motor skills, including graphics. Conduct: cardiac magnetic resonance imaging was requested to assess associated vascular malformations; USG total abdomen to evaluate possible other hemangiomas; increased the dose of phenobarbital to 100mg; awaiting assessment of serum blood level.
- Medical clinic - attended on 06/26/2019, at 1:00 pm, accompanied by the stepmother, without

complaints, already being followed up with the neuropsychiatrist. Clinical evaluation was performed without changes. Conduct: requested laboratory tests and awaiting tests already requested by the neuropsychiatrist. Return after 03 months.

- Dentistry - attended on 06/28/2019, main complaint of the companion "he does not like to brush his teeth, so the situation is bad. You have to take a look". Teenager never went to the dentist not being collaborative with oral hygiene. Patient referred by nursing due to the presence of oral lesions. Plaque accumulation was observed on the surfaces of most of his teeth, with proximal cavities (between neighboring faces) and occlusal ones. The destruction of dental surfaces by the carious process was accentuated in their first molars, which no longer had viable crowns. A panoramic radiograph was performed that did not show any intraosseous lesions. His teeth were composed of deciduous and permanent teeth, some of which were still erupting. There is a need for an adaptation of the oral environment, with the elimination of infectious foci, cleaning of cavities and placement of restorative material. Still, the removal of some teeth, including permanent ones, is necessary, which will require rehabilitation planning to maintain masticatory capacity. It is important to note that the presence of intraoral angiomas is exuberant, including in areas adjacent to the teeth that should be removed. There was an increase in volume in the lower lip, anterior cervical lymph node E, enlarged/painful, firm and mobile consistency. In this first consultation, cleaning and fluoride application were performed. Thus, it is essential to draw up a treatment plan considering the need for interventions in a hospital environment. The patient was rescheduled and must be followed up at the dentistry service.

As academics, it was possible to realize that one of the essential factors for comprehensive care in assisting adolescents with the syndrome was the fact that the health unit, a school clinic, belongs to a private Faculty with a commitment to teaching and has an agreement with the System Unified Health System (SUS), providing a field of practice for the internships of its undergraduate courses, so it was able to meet the demand in an extremely fast time respecting the fundamental principles of SUS: universality, comprehensiveness and equity.

What makes us think that maybe in another service, where it depends exclusively on SUS, these appointments with specialties do not happen with such agility.

Thus, the results of this study point out the great importance of health institutions that have responsibility for teaching/research, collaborating with the training of health professionals for SUS, thus providing an improvement in the quality of life of the assisted population.

DISCUSSION

The literature points out that with regard to Sturge-Weber Syndrome, the symptoms must be addressed separately, since there is no specific treatment⁽¹⁰⁾. Support for the patient with the syndrome involves several issues, including psychosocial ones, since they play an important role in monitoring the patient and their family members. Mainly because he is an adolescent who, for the control of comorbidities, needs the support of his family in order to continue the treatment.

During adolescence it is possible that health is compromised in an irreversible way, so it is up to health professionals to pay more attention at this stage of life, in order to recognize the demands of this population and to intervene in a timely manner, so it is extremely important that adolescents access health services⁽¹²⁾. We realized when following

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this case that because it is a rare disease and, mainly because it affects an adolescent, the need for a different look is evident, since the Sturge-Weber Syndrome presents neurological, cutaneous, ocular and oral manifestations that can (or not) be associated with each other.

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Skin lesions of the syndrome generally do not require early aesthetic therapy, as there is a possibility that this stain may be disguised with the use of cosmetics or be permanently treated by laser therapy⁽¹⁰⁾. Our patient is in a challenging moment in a new school, and his case reveals that living with a change in the pigmentation, texture or shape of the skin can bring consequences, especially in this phase of discoveries, vanity and transformation that is the adolescence.

Ocular complications of the syndrome are manifested, such as glaucoma and vascular malformations of the conjunctiva, episclera, choroid and retina⁽¹⁴⁾. Annual follow-up for glaucoma, using funduscopy and tonometry, is recommended for all patients with Sturge-Weber Syndrome⁽¹⁰⁾. In this case, it was possible for the patient in question to return to eye care, signaling that our performance as academics was of such importance in identifying the need and making the referral after contact with the professional.

The management of Sturge-Weber Syndrome involves multidisciplinary involvement, with the neurologist being one of the important profes-

nals for the early recognition and control of seizures and the prescription of antiepileptic drugs to prevent seizures and, therefore, prevent progressive neurological damage⁽¹⁵⁾. Thus, the outcome of our nursing consultation when referring the patient to the Neuroepidiatrician, who changed the dose of phenobarbital and requested specific imaging tests, gives us the satisfaction of having collaborated with the promotion of this adolescent's health.

Regarding to oral manifestations, they are present in approximately 38% of cases and may involve hemangiomatic lesions on the lip, oral mucosa, gums, tongue and palate⁽¹⁴⁾.

In the case of our patient, a poor maintenance of oral hygiene was observed during dental follow-up, this deficiency is cited as a consequence of the Syndrome due to the slightly challenged cognitive functioning, which can intensify inflammation and gingival hyperplasia⁽¹⁴⁾.

Dental follow-up for adolescents with Sturge-Weber Syndrome is essential and, in some cases, there is a need to perform procedures only in hospital units due to the risk of bleeding. Dental rehabilitation for these patients is a complex process⁽¹⁴⁾, hence the importance of our patient being accompanied to perform dental extractions and remain under periodic evaluation.

It is evident, during our experience report, the need for team work, because the engagement and involvement among professionals from different areas contributes to better assist the adolescent, due to the variety of changes found in the Sturge-Weber Syndrome, therefore, periodic assessment and monitoring of cases by an interdisciplinary team is of fundamental importance⁽¹⁶⁾.

CONCLUSION

The findings in our patient were similar to those mentioned in the li-

terature and the assistance performed according to the notes described. It was possible to show that in order to provide comprehensive care in the care of adolescents with a rare syndrome, such as that of Sturge-Weber, the involvement of a multidisciplinary team is extremely important.

This experience, as nursing students, allowed us to make several contributions due to an integral look at this individual, who will have to live and adapt to the various physical and emotional changes inherent to his pathology. The possibility of having a multidisciplinary team in partnership with the adolescent's nursing service has enabled us to make significant advances in comprehensive care, which is a characteristic of the health unit involved.

This experience report is expected to contribute by helping many professionals during their work process and the academics an integral look at the patient during their learning. ■

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