PROGRESSIVE OSSIFYING FIBRODYSPLASIA IN PEDIATRICS: AN INTEGRATIVE REVIEW FOCUSING ON NURSING

ABSTRACT

Objective: to identify the nursing care/assistance to pediatric patients with FOP in Primary Health Care. Method: integrative review was conducted in October 2020, in five databases and gray literature. With national and international descriptors. The eligibility of studies was performed by independent reviewers. Results: of 1213 potential studies, 2 studies were included in the final sample. Three thematic care directions followed: Nursing contributions to the early recognition of the disease, the importance of health education, and the prevention of injuries. Conclusion: The results obtained added knowledge about the assistance to pediatric patients with FOP. Scientific studies about nursing care for this disease are still scarce, so it is recommended that new studies be developed, given the significance of nursing's role in rare diseases.

Descriptors: Myositis Ossificans; Nursing; Pediatric; Primary Health Care; Review.

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RESUMO


Descritores: Miosite Ossificante; Enfermagem; Pediatria; Atenção Primária à Saúde; Revisão.

RESUMEN

Objetivo: identificar las acciones de atención/asistencia de enfermería a pacientes pediátricos con FOP en la Atención Primaria de Salud. Método: revisión integradora, las búsquedas se realizaron en octubre de 2020, en cinco bases de datos y en la literatura gris, con descriptores nacionales e internacionales. La selección de los estudios fue realizada por revisores independientes. Resultados: mediante una búsqueda amplia se identificaron 1213 estudios y se incluyeron 2 artículos en la muestra final. Se siguieron tres direcciones temáticas de atención: aportes de la enfermería para el reconocimiento temprano de la enfermedad; importancia de la educación para la salud; y prevención de traumatismos. Conclusión: los resultados aportaron conocimiento adicional sobre la atención a pacientes pediátricos con FOP. Los estudios científicos sobre la atención de enfermería a esta enfermedad aún son escasos, por lo que se recomienda desarrollar nuevos estudios, dada la importancia que tiene el papel de la enfermería en las enfermedades raras.

Desciptores: Miositis Osificante; Enfermería; Pediatría; Atención Primaria de Salud; Revisión.

INTRODUCTION

Myositis Ossificans Progressiva, also called and known as Fibrodysplasia Ossificans Progressiva (FOP), is a rare genetic disease characterized by malformation of the first toes of both feet and heterotopic ossification (OH), including connective tissues, muscles, tendons and ligaments.1 Although the term recognized by the Health Sciences Descriptors is Myositis Ossificans, for the purposes of this study the pathology will be presented as Fibrodysplasia Ossificans Progressiva (FOP), since this is the current term that has been used in several scientific studies to refer to pathology.1,2 According to The International Fibrodysplasia Ossificans Progressiva Association2, there are currently 900 known cases of the disease worldwide. Despite its...
low prevalence, about 0.6 - 1.3 cases per million individuals have added space for discussion worldwide due to the highly limiting and disabling condition that it provides to those affected, in a short period of time.

FOP is confused with other pathologies, such as cancer and fibromatosis, due to the lack of knowledge about the disease. The difficulty in completing a diagnosis and the lack of information about this pathology exposes the patient to inadequate treatments that contribute to an early evolution of the clinical picture.³

A clinical study carried out in seven international centers, with the objective of describing the characteristics of FOP and evaluating the progression of the disease, showed that the clinical manifestations of the pathology begin in childhood, resulting in an increase in disabilities and functional limitations throughout life. That said, treatments should target the pediatric population in order to alleviate bone limitations that occur with advancing age.¹

The line of care for rare diseases within the scope of the SUS is described in Ordinance No. 199, of January 30, 2014, of the Ministry of Health (MS), which establishes the National Policy for Comprehensive Care for People with Rare Diseases.⁴ In this scenario, the importance of Primary Health Care (PHC) is highlighted, which is the gateway to the Unified Health System (SUS) and is responsible for monitoring, continuity of clinical follow-up and comprehensive care, as a communication center for the Network of Health Care (RAS).⁴ People with FOP, despite presenting impairments of different proportions, may not require tertiary care.⁵

It is noteworthy that within the PHC, nursing has a leading role. In addition to carrying out activities inherent to their professional practice, in accordance with the legal provisions of the profession, such as consultations, examination requests, medication prescriptions and referral to other services, it contributes to the process of continuing education of the multidisciplinary team, collaborating directly and indirectly to the improvement of assistance.⁶,⁷

However, it is known that FOP is a disease that is little known by health professionals, due to its low incidence, rarity and complexity. Therefore, the development of scientific studies in the form of reviews is justified, with a view to disseminating knowledge, especially among nursing professionals, as an essential tool for the (re)knowledge and diagnostic investigation.
of new cases and, consequently, improvement of the quality of assistance.

Therefore, the objective of this study was to identify nursing care/assistance actions for pediatric patients with Fibrodysplasia Ossificans Progressiva in Primary Health Care.

Materials and methods

This is an integrative review (IR), whose purpose is the synthesis of knowledge and the incorporation of the results of significant studies in clinical practice. The six stages of Ganong were followed, namely: 1) elaboration of the research question 2) search in the literature; 3) data collection 4) analysis of findings; 5) interpretation of results; 6) report of the integrative review.

For the elaboration of the research question, the Population, Concept and Context (PCC) strategy was used, in which the following were defined: P - pediatric patients affected by Fibrodysplasia Ossificans Progressiva; C - nursing care; C - primary care. Based on these definitions, the following guiding question was established: “How has nursing been inserted and/or considered in the provision of care/assistance to pediatric patients with FOP in Primary Health Care?”.

The following inclusion criteria were considered: being indexed in national and international journals, in Portuguese, English and Spanish; understand the population of pediatric patients aged 0 to 21 years with FOP; identify the health care that can be applied by nursing; encompass the context of primary health care. No time limits were defined for this research, due to the low rate of publications found in previous searches in the databases. As exclusion criteria: review articles, duplicates and not available in full.

To survey the articles, the terms indexed by the Health Sciences Descriptor (DeCS) were identified: myositis ossificans; primary nursing; health care; nursing care; primary health care and nursing; and by Medical Subject Headings (MeSH): Myositis Ossificans; Nursing Care; Primary Nursing; Primary Health Care; Delivery of Health Care; Nursing. The descriptors were combined with each other, according to the specifics of each database, using the boggling operators “AND” and “OR” to formulate the search strategy.

The bibliographic survey was carried out with the databases with the support of the Portal of periodicals of the Coordination for the Improvement of Higher Education Personnel (CAFE - CAPES):
PubMed (National Library of Medicine); CINAHL (Cumulative Index to Nursing and Allied Health Literature); Web of Science; Scopus; base Gray literature databases were also consulted: EthOS, Theses Canada, Dart; and CAPES Catalogue.

An example is a search strategy performed in PubMed/MEDLINE with the referred terms and the entry terms:

**TABLE 1** – Example of the search strategy used in the PubMed/MEDLINE database, Curitiba, 2020

<table>
<thead>
<tr>
<th>Order</th>
<th>Search strategy</th>
<th>Studies accessed</th>
</tr>
</thead>
<tbody>
<tr>
<td>#1</td>
<td>“Myositis Ossificans” OR “Fibrodysplasia Ossificans Progressiva” OR “Progressive Myositis Ossificans” OR “Progressive Ossifying Myositis” OR “Myositis Ossificans Progressiva”</td>
<td>2,613</td>
</tr>
<tr>
<td>#2</td>
<td>”Nursing Care” OR ”Care, Nursing” OR ”Management, Nursing Care” OR ”Nursing Care Management” OR Nursing OR Nursings OR ”Primary Nursing” OR “Nursing, Primary” OR “Primary Nursing Care” OR “Care, Primary Nursing” OR “Nursing Care, Primary” OR “Primary Health Care” OR “Care, Primary Health” OR “Health Care, Primary” OR “Primary Healthcare” OR “Healthcare, Primary” OR “Primary Care” OR “Care, Primary” OR ”Delivery of Health Care” OR ”Delivery of Healthcare” OR ”Healthcare Deliveries” OR ”Healthcare Delivery” OR ”Delivery, Healthcare” OR ”Delivery, Healthcare” OR ”Health Care Delivery” OR ”Delivery, Health Care”</td>
<td>1,891,630</td>
</tr>
<tr>
<td>#3</td>
<td>#1 AND #2</td>
<td>21</td>
</tr>
</tbody>
</table>

SOURCE: authors (2020).

Studies were selected by two independent reviewers, according to previously established inclusion and exclusion criteria. It started by excluding duplicate studies. In sequence the studies were read the titles and abstracts and finally, the reading in full. To obtain the data, a table was created with the following items: article order number; title; authors; knowledge area; method; country of publication; journal impact factor; year of publication; main objectives; main results; study recommendations/ conclusions.

The extracted data were transcribed into spreadsheets built with the help of Microsoft OfficeExcell® 2010 and later organized into tables for better visualization of the results. The analysis of the articles included was carried out through narrative synthesis. The review followed the recommendations of the Preferred Reporting Items for Systematic Reviews and Meta-Analyses – PRISMA.11 checklist.
RESULTS

Through the search carried out in the different databases, 1,213 studies were identified, and the inclusion in the final sample comprised two studies that answered the research question. According to the databases and articles identified, Cinahl (n=12), Embase (n=15), PubMed (n=21), Scopus (n=7), and Web of Science (n=1,113), CAPES Catalog (n=35), DART (n=4), and EthOS (n=6). No studies were found in These Canada. The process of including articles for this review is shown in Figure 1.

FIGURE 1– Flowchart of the process of identification, selection, eligibility and inclusion of studies.

Among the articles included, both are from the field of medicine, published in Brazil and Spain, in 2013. The main characteristics extracted from the articles are presented in Table 2. And, the objectives, main results and, the main recommendations and/or conclusions are shown in Table 3.

**TABLE 2– Characterization of scientific productions on FOP, Curitiba, 2020**

<table>
<thead>
<tr>
<th>N.</th>
<th>Title</th>
<th>authors</th>
<th>Journal title</th>
<th>Research method</th>
<th>Impact factor</th>
</tr>
</thead>
<tbody>
<tr>
<td>A1</td>
<td>Fibrodisplasia ossificante progressiva: diagnóstico na atenção primária</td>
<td>Garcia-Pinzas,J Wong,JEB Fernández,MAP Rojas-Espinoza, MA</td>
<td>Rev. Paul. pediatria</td>
<td>Case report</td>
<td>0.1232</td>
</tr>
<tr>
<td>A2</td>
<td>Hay lugar em atención primaria para las enfermedades poco frecuentes? El caso de la fibrodisplasia osificante progressiva</td>
<td>Morales-Piga,A Ribes,MG Álvaro,PA Álvaro,CC La Paz,MP de Bachiller-Corral,J</td>
<td>Atención primaria</td>
<td>Case report</td>
<td>1.087</td>
</tr>
</tbody>
</table>

TABLE 3-Synthesis of articles included according to objectives and main results, recommendations and conclusions of the study, Curitiba, 2020

<table>
<thead>
<tr>
<th>N.</th>
<th>Main objectives</th>
<th>Main results</th>
<th>Recommendations / Conclusions</th>
</tr>
</thead>
<tbody>
<tr>
<td>A1</td>
<td>To demonstrate that it is possible to diagnose Fibrodysplasia Ossificans Progressiva in primary care.</td>
<td>The age at diagnosis was 10 years. Presence of bilateral hallux valgus since birth. Onset of the first symptoms at 4 years of age, with the appearance of painful and mobile tumors and, later, tumors of hard consistency, immobile and non-painful, without inflammatory signs. Some OH were related to traumatic antecedents. There were movement limitations with elbow and spine stiffness. Non-steroidal anti-inflammatory drugs and corticosteroids were used in the treatment.</td>
<td>The authors consider it possible to diagnose FOP in primary care. They recommend carrying out a clinical evaluation and highlight the importance of intervening in the development of the disease, thus preventing its aggravation.</td>
</tr>
<tr>
<td>A2</td>
<td>Assess the population of patients with FOP in Spain*</td>
<td>The population evaluated was 24 people. The mean age at diagnosis was 7 years, with a mean delay of 3 years (from the onset of the first symptoms). Of the patients: 21 had congenital bilateral hallux valgus and all had endochondral ossification; half of the cases reported having suffered muscle trauma prior to bone injuries; 12 patients had functional limitations*</td>
<td>The authors point out that care for people with rare diseases can be improved. Therefore, they recommend a systematic approach from the perspective of care in primary care and coordination in a care network with other secondary and/or tertiary services*</td>
</tr>
</tbody>
</table>

NOTE: * Free translation by the authors.
SOURCE: Authors (2020).

DISCUSSION

The results obtained in this integrative review allowed the identification of three thematic care directions: nursing contributions in the early recognition of FOP; the importance of health education; and, the prevention of injuries.

Nursing contributions in the early recognition of FOP

Rare diseases (RD) have a high degree of complexity, and most of them present peculiar signs and symptoms essential to restrict and conclude a diagnosis. When dealing with FOP, the results of this study point to important characteristics that should be observed in this disease – bilateral congenital hallux valgus and OH. Likewise, a review study that aimed to present the pathophysiology and clinical characteristics of FOP showed deformities in the first toes of both feet, at birth, and OH, with important clinical findings present in all cases.

In view of the above, the anamnesis and physical examination emerge as instrumental strategies of great importance...
for the identification of RD, such as FOP, since they allow obtaining the necessary subsidies for the correct diagnosis and adequate care planning.\textsuperscript{5,14} Professional care

The nursing care system is scientifically based on the Systematization of Nursing Care (SAE), a methodology that organizes care in a systematic way, with a view to improving the quality of care. The development of SAE is based on clinical reasoning developed from the five stages of the Nursing Process (NP): Data collection and investigation; Nursing Diagnosis; Assistance Planning; Implementation of assistance; and, Evaluation of results. Anamnesis and physical examination are components of the first stage of the NP.\textsuperscript{14,15}

Nursing assumes a leadership role in the scope of primary care, developing various care and management activities. Among them, the Nursing Consultation stands out, a space for the development of clinical practice, in which interaction between patient and professional is possible, and space is opened for interventions such as prescriptions, guidelines and referrals, according to the needs of each patient and, based on the PE.\textsuperscript{15,16}

FOP is a congenital genetic disease, which presents its first symptoms in the first decade of life, around 4 years of age, as observed in the present study. There is the nursing consultation in childcare, to assess child growth and development, as a valuable tool in the early detection of FOP signs, since from there the nurse can detect health problems and intervene early.\textsuperscript{4,17}

**The importance of health education**

The present study identified a mean age of more than seven years for the diagnosis of FOP, coinciding with Sferopoulos et al.\textsuperscript{18} who showed a mean age of 9 years. Late diagnosis can be justified by the vast knowledge gap on the part of health professionals in relation to rare genetic diseases, either because of their rarity, the superficial coverage of this subject during academic training, or because they are little explored in the context of public health.\textsuperscript{19}

It is known that education is a continuous process of knowledge creation. Thus, health education is an important ally for the recognition of rare pathologies little discussed in the field of health in general, since it provides training, updating professional practice and, consequently, improving care for patients with these diseases.\textsuperscript{7}

Regarding nursing, in addition to being care professionals, they also play the
role of educators, especially in the context of primary care, contributing, participating and carrying out activities inherent to health education, in order to mediate the exchange of experiences, knowledge and reflection of work processes among the multidisciplinary team.7,16

Despite its importance, disseminating technical-scientific knowledge about RD is not enough to guarantee adequate assistance. Society, together with the managers of the health system, need to see this public as members of the community, in order to reduce the existence of the invisibility of these patients, thus ensuring full access to health.12

Trauma prevention

The articles analyzed in this review pointed to traumas prior to ossifications. Intramuscular injections, falls, biopsies and other soft tissue trauma are important triggers for new episodes of flare-ups – symptoms of active FOP, as they can stimulate the wrong bone formation. Because OH is easily confused with cancerous nodules, biopsy is the most common trauma for people with FOP, and therefore, like the others, it should be avoided.20

It is known that the evolution of FOP is associated with the emergence of numerous limitations, resulting from OH that somehow interfere with the individual's quality of life.1,3 Akyuz13 in his study of FOP characterization related traumas as surgical interventions and invasive procedures, which occur as the bone lesions appear.

The limitations of movement, imposed by the wrong bone formations, characteristic of FOP, affect mainly areas of the body such as the spine, hip and peripheral joints, promoting, over time, an increasing decrease in the mobility of those affected. These limitations were observed in the results of the present review, compromising the individual's autonomy and independence as the disease progresses. In this sense, the prevention of soft tissue trauma is a priority in the management of FOP.1

The prevention of flare-ups, which lead to OH, involves recognizing and mastering the disease, in order to avoid actions that stimulate ossification, such as intramuscular injections, falls, biopsies, excessive muscle stretching, surgical procedures and viral diseases.20 Finally, since the treatment of FOP is only symptomatic, with the use of non-steroidal anti-inflammatory drugs (NSAIDs) and corticosteroids, which aim to alleviate pain
and prevent flare-ups, the multidisciplinary team must be attentive to the recognition of these patients, for the prevention of damage, since the progression of the disease leads to progressive and severe limitations that will lead the individual to a shorter useful life\(^1\),\(^2\), to an early family and social loss, potentially preventable.

As a limitation for the development of this study, the scarcity of scientific production on FOP in the nursing area is highlighted, especially with a focus on the pediatric population.

**CONCLUSION**

The data from this study favored greater knowledge about care for patients with Fibrodysplasia Ossificans Progressiva within the scope of Primary Health Care. As well as the knowledge of some of the peculiarities and needs of patients affected by this disease, which were discussed from the perspective of the contribution of nursing to the recognition of FOP, the importance of health education and the prevention of trauma.

So it can be seen that nursing plays a relevant role in the care of pediatric patients with FOP in the discussed axes. It was found that the main care needs for these patients are centered on early diagnosis and damage prevention. In view of this, it is also worth highlighting the importance of the performance of a qualified multidisciplinary team in order to achieve the best and timely care for this public and their family, from the perspective of integrality and access to health.

The existence of a knowledge gap in this area is highlighted, and it is suggested that new research be developed on nursing care for patients with FOP, and also, concerning the quality of life of these patients and their families, in order to be produced knowledge about the peculiarities and needs of care.

**Conflicts of interest:** The authors declare that there is no conflict of interest.

**REFERENCES**


