The Xeroderma pigmentosum syndrome: an experience of integral attention to children with special needs in Cuba

El síndrome Xeroderma pigmentosa: una experiencia de atención integral a los niños con necesidades especiales en Cuba

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Abstract

A considerable part of the bibliography at present refers to the clinical-genetic description of the Xeroderma pigmentosum syndrome as well as its consequences for the subjects that suffer it, in other cases it is dedicated to present sophisticated formulas to protect people from the ultraviolet rays, there are very few references to the needs that in the psychological, preventive and educational order emanate from the so-called “infants of the moon”. In order to contribute to solving this problem, a project was developed to improve education for the prevention and comprehensive educational care of children, adolescents and young people of school age who suffer from this syndrome. This article presents the dimensions and indicators defined in the research process to characterize the comprehensive educational care demanded by patients who, in different personal, family and social conditions, face the challenge of living with this peculiar condition.

Keywords: Xeroderma pigmentosum syndrome; comprehensive care; characterization

Resumen

Una parte considerable de la bibliografía en la actualidad se remite a la descripción clínico-genética del síndrome Xeroderma pigmentosa así como sus consecuencias para los sujetos que la padecen, en otros casos se dedica a presentar fórmulas sofisticadas para proteger a las personas de los rayos ultravioletas, son muy escasas las referencias a las necesidades que en el orden psicológico, preventivo y educativo emana de los llamados “infantes de la luna”. Con el propósito de contribuir a resolver esta problemática se desarrolló un proyecto de mejoramiento educativo para la prevención y atención educativa integral a niños, adolescentes y jóvenes en edad escolar que padecen este síndrome. En el presente artículo se presentan las dimensiones e indicadores que se definieron en el proceso investigativo para realizar la caracterización de la atención educativa integral que demandan los pacientes, que en disímiles condiciones personales, familiares y sociales se enfrentan al desafío de convivir con este peculiar padecimiento.

Palabras clave: síndrome Xeroderma pigmentosa; atención integral; caracterización

Introduction

The Xeroderma pigmentosum syndrome (XP) was first described as far back as 1874 by doctors Ferdinand Von Hebra and Moritz Kaposi[1], but it is in recent years that a greater number of references have been found to this rare genetic disease whose name results from the union of the terms: xeroderma (dry skin) and pigmentosa in allusion to the characteristic pigmented lesions of the skin.

The bibliographic reviews carried out[2,3,4] emphasize the clinical aspects of this disease in terms of etiology, symptoms, characteristics, differential diagnosis and genetic

traits, among others. Other sources[5,6] refer to the search for effective therapies, some of which are sophisticated and expensive, and would therefore be unattainable for people living in developing countries.

The psychological, emotional and educational needs of the so-called "infants or children of the moon" are not widely discussed in the literature[7,8]. Around this problem multiple questions arise: how does a child or adolescent assume the fact of having been diagnosed with this syndrome; what role does the family play in this situation; how can their interpersonal relationships be strengthened in the very particular life regime imposed by this diagnosis; how can the educational attention they require be guaranteed in the new conditions; these are some of the questions to which answers must be found in order to guarantee the integral attention that favours their harmonious formation.

For this purpose, a project was developed to improve education for the prevention and comprehensive educational care of children, adolescents and young people of school age who suffer from this syndrome. The present article presents the dimensions and indicators defined in the research process to characterize the comprehensive educational care demanded by patients who, in different personal, family and social conditions, face the challenge of living with this peculiar condition.

Methods

The Xeroderma pigmentosum Syndrome (XP). Main considerations

The numerous clinical studies of this pathology agree on the following main common features:

- It is a rare genetic condition that translates cellular hypersensitivity to ultraviolet radiation in association with abnormal deoxyribonucleic acid repair, producing freckling, photophobia and subsequent neoplastic changes in sun-exposed areas[9].
- is a genetic DNA repair disorder in which the body's normal ability to remove damage caused by ultraviolet (UV) light is impaired[10].
- Hereditary inability of the skin to repair DNA damage from UV light[11].
- is an autosomal recessive genodermatosis that mainly affects skin, eyes, and brain; it is characterized by pigmentation, malignant neoplasms, and sometimes involvement of other organs[12].

For Capote Mira[13], Xeroderma pigmentosum is a genetic disease that is inherited in an autosomal recessive pattern, which means that the affected individual is homozygous for the mutation responsible for the syndrome; it is a mutation that affects the DNA repair system in the NER fraction (excision - nucleotide repair), which takes place by exposure to ultraviolet light and chemically induced damage to skin cells.

The difference with people who do not have this genetic defect is that the latter have an enzymatic team specifically for endonucleases that is integrated in the repair functions of the damage that occurs in the cellular DNA when there is exposure to ultraviolet (UV) light and to a lesser extent to other harmful agents such as drugs and chemical carcinogens. These damages accumulate and perpetuate the mutations to the DNA that will finally be reflected in the development of a malignant neoplasm. It occurs in both sexes and in all races and has often been seen to be inbred between parents[13].

Individuals who suffer from the disease and who are homozygous for the mutation of the gene that encodes for this type of protein cannot repair the damage to the DNA of the skin cells by the action of ultraviolet rays and their unprotected skin when it receives sunlight becomes red, blistered, irritated and in these regions there is a mark in the form of freckles and spots in the most exposed areas, first in an isolated form and then converging in larger lesions and tending to become tumours in their three fundamental forms: basal cell carcinoma, spinocell carcinoma and melanoma[14].

Xeroderma pigmentosum syndrome is accompanied by eye symptoms such as photophobia, tearing, serous or mucopurulent discharge and reflex blepharospasm. Eight types of Xeroderma pigmentosum are recognized, which imply different levels of affectation to the health of the subject who suffers from it, they have been named with letters from A to G and the last one is called the variant form, each type is characterized by a different genetic change in the DNA repair system[15].

Sanctis-Cacchione syndrome is the most severe form of Xeroderma pigmentosum and involves skin and eye manifestations with severe neurological and somatic disorders, resulting in lethal damage. Its main clinical features include: progressive deterioration of the nervous system with convulsions, ataxia, spasticity and incoordination, hyporeflexia or areflexia, microcephaly, moderate or significant growth disorders, mental retardation, neurosensory deafness, immature sexual development, low intelligence and skin lesions in exposed regions[16,17].

There is still no curative treatment for XP Syndrome, therapies such as gene proteins are being tried, so treatment is currently oriented towards prevention, that is, avoiding exposure to solar radiation, as well as the effects caused by free radicals[18] Affected patients should avoid exposure to the sun's rays. This forces the patient to change his or her entire behaviour. In addition, they must avoid all known carcinogens. XP patients have to go outdoors with gloves, hat, protective clothing such as light-colored,

tightly woven clothing, and sunglasses, which makes it difficult to live a normal life.

Encourage the darkening of rooms; the windows of their houses must have special ultraviolet ray glasses, restriction of activity during the day and at night without fluorescent lights, periodic medical examinations, and early removal of any suspicious lesions/tumors. The increasing presence of this syndrome in our environment[19] has posed a challenge to the authorities of the agencies, ministries, institutions and organizations that must attend to the people afflicted, which is why an educational improvement project was developed in the province of Villa Clara, since it is the territory with the most diagnosed cases in the country[20].

Most of the patients diagnosed are of school age and are cared for by a multidisciplinary team made up of dermatologists, neurologists, ophthalmologists and psychologists.

The Education Improvement Project creates the conditions for an individualized, contextualized psycho-pedagogical characterization that specifically addresses the special educational needs of each of the schoolchildren, as a premise for implementing actions aimed at providing them with comprehensive care.

With an approach aimed at preventing the appearance of secondary defects associated with the disease, such as: visual, motor and mental disorders, depression, anxiety, low self-esteem, among others[20,4]. The aim of Cuban education is based on equal opportunities for all its students. Everyone can succeed, learn, develop, access knowledge and culture, use their intellectual and practical skills and reach the maximum development of their abilities, whether or not they can attend an educational institution.

Development and discussion

In the Cuban educational context, the educational attention is conceived as a system of planned actions, where the aids are established through the formats of joint action, personalizing the facilitating instruments and qualifying the context from the role of the educational agents in their opportunity as mediators to contribute to human development[21].

The authors of this article consider that educational care should integrate the action of all those factors and agents of socialization that contribute to the personal formation of children, adolescents and young people, but it is up to the school as an institution and its teachers and directors to study in depth and interpret each situation and to make available the resources to provide the necessary responses in a timely manner.

Children with an illness that prevents them from attending school live in a world with psychological and social barriers that make it impossible for them to get to know their surroundings; they cannot continue to carry out the activities of their daily lives, relate to their peers, their habits and the rhythm of their lives are altered and changes in their behaviour may occur.

The search for resources, help and support is very necessary to provide effective care for these schoolchildren. The pedagogical work, aimed at teaching, educating and working for the integral personal development of the students must be a process of permanent search for methods, procedures and means that can guarantee maximum efficiency and quality in the fulfillment of the missions, objectives or general purposes, without relegating or segregating anyone[22].

This permanent search must start with an adequate diagnostic process, which, as a pedagogical process, allows us to know the individual and group risk factors of the schoolchildren, families and communities; the real particularities of the students, the potentialities of both the students and their different life environments: school, family and community and to cover in a general way the state of the educational attention that a certain group or category of schoolchildren receive[23,24].

In the question that concerns us, it is necessary to distinguish between the differential clinical diagnosis that the minors already possess after a rigorous clinical-genetic study that is offered by the health institutions and the diagnosis of the educational care they receive that constitutes the purpose to be presented in this article.

It is necessary to diagnose the work done by all educational and socialization[25] factors that influence in some way the integral formation of school children with XP, so specific instruments are used for each one.

To this end, it was necessary to specify the dimensions and indicators that would guide the research work of the project in the elaboration of a battery of diagnostic instruments made available to the professionals involved in the education of these schoolchildren.

Several steps were followed to prepare the battery of diagnostic instruments:

1st- Deepening in the bibliography and the study of this syndrome and the international vision on the attention to the people who suffer from it.

2nd- Analysis of documents: the characterizations carried out on the minors in the sample by the social workers in charge of their care and the database prepared by them and the psycho-pedagogical characterizations carried out by the teachers who attend them were analyzed.

3rd- Interviews: applied to 16 schoolchildren (representing 51.6% of the sample); relatives; teachers who serve them; social workers; officials and managers of the CDO, MINED and the Dermatology Specialist who heads the multidisciplinary team that provides clinical care to minors.

4th- Triangulation of sources: The information obtained from the bibliographic review and the consulted documents was related, as well as that provided by the

sick school children themselves, their relatives, doctors, officials, managers and social workers.

This allowed us to verify that there are not enough psycho-pedagogical sources of information, which serve as the basis for the educational care of these minors; that the psycho-pedagogical characterizations carried out by the teachers who attend them, only address isolated elements, do not reflect their true needs and potential; There is insufficient knowledge of the clinical and psycho-pedagogical characteristics of the staff and managers responsible for caring for these minors.

In response to these regularities, through work tables (in which it was necessary to carry out several rounds), the researchers defined the following dimensions and indicators to carry out an objective characterization of educational care for schoolchildren with XP syndrome, which responds to their needs and potentials.

The proposed Dimension I is based on the study of the personal factors of each child and the indicators are expressed below:

- Age and time of appearance of the disease: this refers to the age of the schoolchild at the time of investigation and the time that has elapsed since the symptoms of the disease appeared. The phase of acceptance or rejection of the diagnosis and the awareness of living with the disease will depend on this.

- How the diagnosis is assumed: if the fact of having a positive diagnosis of this disease is a reason for sadness, depression, pessimism on the part of the child or on the contrary there is a positive and optimistic assumption of the diagnosis expressed in the care of his or her health.

- Level of knowledge about the illness: according to the age of the schoolchild, he/she should know the essential elements of the illness and the need to take care of his/her health.

- Level of satisfaction with teaching activities: it inquires into the degree of satisfaction that schoolchildren have with the work carried out by teachers and other professionals: physical culture, library activities, and computer science.

- Level of satisfaction with pioneer, sports and artistic-cultural activities: this refers to whether schoolchildren are satisfied with these types of activities, which should be integrated based on their specific characteristics, organized in timetables and spaces so that they can be incorporated.

- State of interpersonal relations with peers: how communication with peers is encouraged and should be stimulated in children from the moment they join the family group, enhancing their expressive and communicative abilities with others, especially with peers with whom they share interests and points of view.

The proposed Dimension II is based on the study of the Factors of the family context and the following indicators:

- Number of persons living with the child and their degree of kinship: this will make it possible to determine whether the family is nuclear, extended or extended, the composition of the family nucleus, who is in charge of the child's education, the presence of the paternal or maternal figure, and relevant aspects for the care of the child.

- Quality of intra-family relations: In family decision-making, one of the members of the couple should not be predominant; decisions should be taken by mutual agreement, and the rest of the family members should also participate. Each member of the family nucleus should have age-dependent responsibilities.

- Level of knowledge about the disease: expresses the domain that the family should have over the essential elements of the disease, especially to ensure the health care of their child.

- Level of risk perception about other pathologies that may accompany the syndrome: it expresses the degree of awareness that the family has about the possibility that their child suffers from other disorders associated with the disease, especially when they do not take adequate care of their health.

- Mastery of the evolution of your child's disease: this refers to the stage of the disease and the possible prognosis of the disease, knowledge of which allows the family to prevent the aggravation of the disease and the appearance of other associated disorders.

- Degree of satisfaction with the educational care received by your child: it inquires into the degree of satisfaction that the relatives have with the work carried out by teachers and other professionals: physical culture, library and computer activities.

The factors related to the school context are expressed as dimension III and the indicators are shown as follows:

- Existence of teachers and professors to attend to minors with Xeroderma pigmentosum syndrome of school age: they must have pedagogical experience and correspond to the level and type of education in which the schoolchild with the syndrome is found.

- Experience of teachers in working with these children: check whether the teachers already have experience in working with this type of schoolchild.

- The level of knowledge of municipal managers and officials regarding the care provided to children with the syndrome: check whether managers and officials of education at different levels are aware of the care provided to schoolchildren with the syndrome.

- Level of knowledge and control exercised over actions

for the educational care of minors: checks whether control and follow-up are exercised over the care provided to schoolchildren

- Level of knowledge about the illness of minors: this expresses the mastery that teachers and professors must have over the essential elements of the illness and the health care of the child who suffers it.

- Mastery of the psycho-pedagogical characterization of their schoolchildren: it analyses whether the teachers and professors in charge of this task possess the relevant level of knowledge and skills to carry out the psycho-pedagogical characterization of their schoolchildren with a personalized and developmental approach.

- Level of preparation received by teachers for the educational care of their students: Find out if the school's methodological work includes actions aimed at preparing teachers and other specialists in educational care of these students, including compliance with curricular strategies related to education for health and sexuality, environmental education for sustainable development, among others.

- Degree of satisfaction of teachers with the preparation received in order to provide educational care for these children: it expresses whether teachers and professors are prepared to carry out their educational work and are satisfied with it.

- Existence of preventive educational strategies in the educational care of children with the syndrome: it specifies whether the teachers in charge of the teaching-educational process develop preventive actions coordinated with the family and the community where their schoolchildren live.

Finally the dimension IV declared as the factors of the community context the indicators listed below have been selected:

- Community structure with the presence of community leaders: check if the positions and leaderships of the community are present and all the responsibilities are covered.

- Level of knowledge about the disease: it expresses the mastery that community socializing agents should have over the essential elements of the disease and the health care of the child who suffers from it.

- Level of information about the existence of children with the syndrome in their area of action: community agents must be aware of the existence of children with this syndrome in their area of action in order to make their resources available for educational care of these children.

- Relationship with the institutions that provide care for these minors: community agents should establish close interrelationships with other educational institutions involved in the care of minors, especially the family, the school and the cultural centres in their area.

- Planning of training activities for community agents on the treatment of minors: find out about the existence of activities carried out for the training of community agents, their frequency and effectiveness.

- Planning of actions aimed at the educational care of minors with the syndrome: this is aimed at ascertaining what type of activities and actions are planned for the educational care of these minors.

- Quality of community actions aimed at the educational care of minors: to evaluate the quality, relevance and results of these actions.

- Ways in which community actions are controlled: to verify the systemic nature and the control exercised to follow up on community actions aimed at providing educational care for schoolchildren.

- Potential of the community to carry out activities that contribute to the educational care of minors: aimed at examining the possibilities that exist in the community to plan activities in accordance with the needs of these minors.

The definition of the dimensions and indicators allowed the characterization of comprehensive educational care for schoolchildren with Xeroderma pigmentosa syndrome and determine the regularities and needs present in them, which enabled the development of guidelines, educational strategies, programs and other teaching materials that are applied in Cuba through a National Project that coordinated by the State and the Government provides priority attention to children, adolescents and young people diagnosed with this condition.

Conclusions

The patients with XP require a multidisciplinary approach, for those who are in school age, it is very important the presence of the professionals of the Education, in charge of conducting their integral formation.

The dimensions and indicators defined allow us to objectively characterize the quality of educational care received by school children with Xeroderma pigmentosum syndrome and to trace preventive educational strategies and actions aimed at solving the needs detected, favoring the harmonious development of school children and their families. The comprehensive educational care provided to minors with this syndrome is a guarantee of a better quality of life and helps to reduce the impact of the syndrome on those affected and their families.

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