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ORIGINAL ARTICLE

Cri du chat syndrome and inclusive education

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Keywords:
Cri Du Chat Syndrome, Social Inclusion, Learning.

Abstract

This study offers reflections on the inclusion of children with Cri Du Chat Syndrome (CCS), a rare condition of genetic origin. **Objectives:** To discuss the inclusion of children with CCS in schools and describe the characteristics, diagnosis, treatment, and main symptoms of the condition, along with the main difficulties students with CCS might face; to assess the knowledge teachers have about CCS and potential teaching resources they may use. **Method:** A digital survey questionnaire was used in this qualitative exploratory descriptive study. **Results:** Only 14.8% of the 27 teachers who responded the questionnaire had knowledge about CCS and had worked in a classroom with adapted pedagogical practices. **Conclusion:** A very small portion of teachers had knowledge about CCS and most had never been in contact with individuals with the syndrome, either in a classroom or during training. A path has to be developed for the inclusion of children with CCS, through a process based on knowledge, affection, love, and respect for differences.

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INTRODUCTION

Cri Du Chat Syndrome (CCS) is a genetic disorder associated with deletions of various sizes in the short arm of chromosome 5 (karyotype 46, XX/XY, 5p).

The syndrome was named after the characteristic cat-like cry of affected children.

Although it is a condition of low prevalence, the alterations/consequences ensuing from it can be quite serious, ranging from death in childhood due to malformation to significant impairment affecting various aspects of the lives of children with CCS.

CCS is a rare condition that causes impairment in multiple areas, including learning. Little is known about how children with the syndrome might develop in a regular classroom environment.

Since the inclusion of individuals with CCS in regular schools is a reality, this study aimed to gather information about the condition and its impact on school life.

Teachers and educators have discussed the importance of inclusive education and sought to acquire specific knowledge about the diseases, anomalies, and difficulties that may compromise a child's ability to learn and interfere with the teaching-learning process.

Children with chromosomal abnormalities in general and individuals with CCS more specifically may present with learning difficulties on account of the several alterations these conditions impose on them, such as microcephaly and mental retardation.

This article offers reflections on the inclusion of children with CCS in regular classrooms based on a review of the literature and insight from a questionnaire answered by teachers designed to find out more about how much they knew about CCS and the characteristics, diagnosis, treatment, main symptoms, and difficulties the condition poses to the development of affected students.

One of the purposes of this article was to help educators deal with the limitations students with CCS face by describing the characteristics of the syndrome and its repercussions on school life.

DEVELOPMENT

Cri Du Chat Syndrome (CCS) was first described by French geneticist Jérôme Lejeune in 1963. The condition described by Lejeune was called the "call of the cat" syndrome on account of the cat-like sound babies with CCS emitted when they cried. Abnormal development of the larynx, one of the several symptoms tied to the condition, is the culprit[1,2].

CCS occurs due to anomalous development of chromosome 5p, which loses a part of its short arm. The syndrome causes a series of alterations that include microcephaly, poor growth, facial abnormalities, mental retardation, malformation of the larynx, widely-spaced eyes, hypotonia, inner corner of the eyes higher than the outer corner, epicanthal folds, poorly formed low set ears, long fingers, single fold in the palm of the hands, neuromotor retardation, and low birth weight. Its main characteristic at birth is the presence of a cat-like cry, as mentioned above. Diagnosis in older patients can be more difficult, since the cry becomes less characteristic as the child grows[2,3,4].

According to some authors, chromosomal alterations are present in 8.1% of fetuses, are one of the main causes of fetal death, and cause 50% of miscarriages[5].

Individuals with CCS are diagnosed via karyotyping, a test performed on a blood sample of the patient. The karyotype of an individual with suspected CCS will show a deletion (loss) of the short arm of chromosome 5; this deletion indicates the presence of CCS with certainty. One in 50,000 children is born with CCS[3,6].

According to the literature, some children with CCS may have difficulty controlling their physiological needs and present behavioral issues such as hyperactivity, excessive head swaying, obsessive or aggressive behavior[6,7,2].

A cat-like cry, poor growth, microcephaly, and mental retardation are present in most cases of the disease. It is also common for children with CCS to develop heart and respiratory problems and significant muscle hypotonia. Other symptoms such as dysfunctional sucking, chewing, and digestive and/or bowel disorders are usually present, as well as significant delayed speech development[2,7].

Unfortunately, there is no treatment or cure for CCS. Individualized follow-up is required for subjects with the disease. Although individuals with CCS share some characteristics, symptoms and degree of impairment vary between patients. Therefore, each individual requires a different approach, depending on their specific needs. In general, several professionals are involved in the care of individuals with CCS, including occupational therapists, speech therapists, physical therapists, psychologists, psychopedagogues, physicians, and dentists[1].

Education is an extremely relevant element in the care provided to individuals with CCS. Developing independence along with communication, socialization, and motor skills improves patient outcomes. Education is a basic right of children and adolescents enshrined in the Brazilian Constitution of 1988, along with the right to wellbeing, health, and plurality, along with provisions against discrimination[8].

In the same way, the Law of Directives and Bases for National Education (LDBEN) (1996) stipulates that schools cannot refuse students with special needs, regardless of disability[9].

Education is an artificial domain of the processes arising from development. It not only influences these processes, but also restructures behavioral functions in their entirety; therefore, speech, writing, gestures, etc. present in education are cultural elements of interaction that constitute human beings[10].

Promoting integral inclusive education relies on the implementation of a society that is also inclusive, a society that naturally allows the existence, free expression, and productivity of people with special needs, whether in terms of educational, personal and/or social endeavors[6].

The inclusion of children with learning disabilities in regular schools, although guaranteed by law, still presents many challenges. It is important that society, educators, and schools enforce the right to education for all and provide for the assistance services needed for the success of inclusive education. According to the Law of Directives and Bases of National Education (LDBEN no. 9394/96), there is much to be done regarding curriculum organization, methodology, evaluation, and inclusion in regular schools. However, the Chapter III, Article 4, item III of the LDBEN stipulates that it is the State's duty to guarantee free specialized educational services to students with special needs, preferably in regular schools[9].

Inclusion is materializing in the public school system. Regardless of the type of disability or degree of impairment, students with special needs attend school in regular classrooms[11]. However, for inclusion to occur and solidify, it is essential that teachers are trained and prepared. Article 59, item III, of the LDB (1996) states that the school system must ensure that students with special needs "have teachers with appropriate training on specialized services who are able to integrate students with special needs into regular classrooms"[9].

Families are an extremely important stakeholder in the education and development of children with special needs. According to Paulo Freire, "there is no education without love [...] and those unable to love unfinished beings cannot educate [...]". Families also need to accept and help in the development of children with CCS[3].

Fiamenghi Jr. and Messa, wrote the following in a study addressing the relationships of families with children with disabilities:

The family is a social force that affects the determination of human behavior and the formation of personality. A family may be defined as a significant social unit embedded in the immediate community and in a broader society. A family is interdependent, that is, the relationships established between family members affect one another and changes in these relationships affect each family member individually or the family system as a whole[12].

Families and schools are key elements in the constitution of individuals, their self-esteem and ability to express emotions and feelings, and other attributes that build bonds of affection and education, particularly for children with special needs[13]. However, it is not enough to include a child with CCS or other disabilities in school without a plan, without knowing their specificities, culture, or the context around them. It is essential to harness the affection, commitment, and training of all everyone involved (schools, teachers, and families). Inclusive education involves the development of a political, social, economic, historical, and pedagogical system around it[14].

METHOD

The references included in this qualitative and exploratory descriptive study included a questionnaire and a literature review including books, scientific articles, documents, monographs, dissertations, theses, and websites of institutions.

The search for literature on the subject included the following keywords: learning; inclusive education; Cri Du Chat Syndrome.

Literature references were collected in a non-systematic manner (scientific articles, case studies, etc.) in the period from January 2020 to October 2020. The scientific databases used in the search for literature were Medline, Lilacs, Scielo and Google Scholar.

Articles with titles or abstracts containing the keywords used in the search for literature and with objectives similar to the ones stated in this article were included. Papers appearing in duplicity in more than one of the scientific databases were excluded.

A questionnaire developed in Google Forms was sent to teachers involved in elementary education. Participation was optional and respondents were anonymized. The questions assessed how much the respondents knew about CCS and whether they had worked with children with the syndrome in a classroom. The questionnaire had four multiple choice and four open-ended questions (Appendix 1), and was made available for answers between August 5, 2020 and October 9, 2020.

ETHICS

The invitation to join the study was first sent to the school principals. The schools that accepted the invitation were sent an e-mail with a link to the questionnaire along with an informed consent form, in which participants joining the study authorized the publication of their answers under the condition that they were used for scientific purposes only and respondents and the institutions at which they worked were anonymized.

In the event of data loss and exposure of the answers submitted via the questionnaire form, participants were promised full assistance free of charge. The contents of the questionnaire were stored in a safe cloud server, with access to such content given only to the authors of the article as stipulated in the informed consent form.

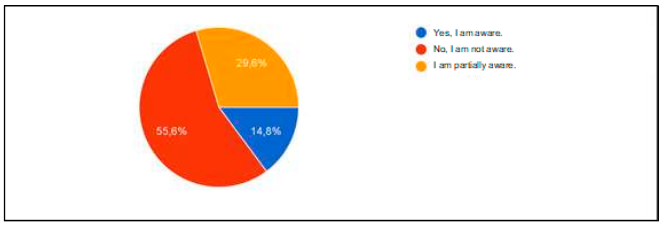
This study was approved by the Research Ethics Committee and given certificate no. CAAE 54461521.1.0000.5415.

RESULTS

The link to the questionnaire was sent to four municipal schools in Northwestern São Paulo, in the period from August 5 to October 9, 2020. The schools were closed at the time due to the COVID-19 pandemic. The teachers were familiarized with Google Forms, which made it easier for them to answer the study questionnaire. The link to the questionnaire was sent to them with the help of school coordinators. The data collected were organized in graphs to facilitate the organization and analysis of the results. Twenty-seven teachers opted to participate in the study.

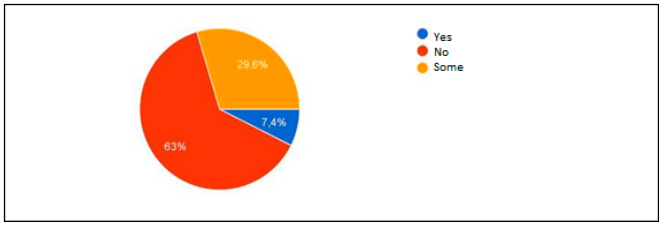
The questionnaire had a total of eight questions, four multiple-choice and four open-ended. The charts shown below depict the answers given to the four multiple-choice questions.

A small portion of the teachers (14.8%) had knowledge of the difficulties children with CCS might experience in their development, while 55.6% had no information about it (Graph 1).



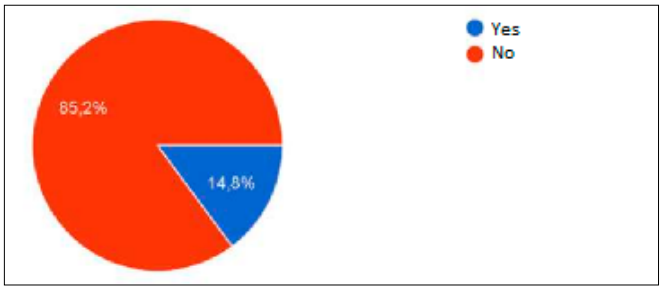
Graphic 1. Awareness of the difficulties children with CCS might experience in their development. Source: Questionnaires answered from Aug. 5, 2020 to Oct. 9, 2020.

When asked about whether they were trained while attending education school on how to teach children with genetic conditions including Down syndrome, Edwards' syndrome, Klinefelter syndrome, and CCS, 63% said they were never taught about it and only 7.4% said they had access to information on how to work with children with genetic syndromes (Graph 2).



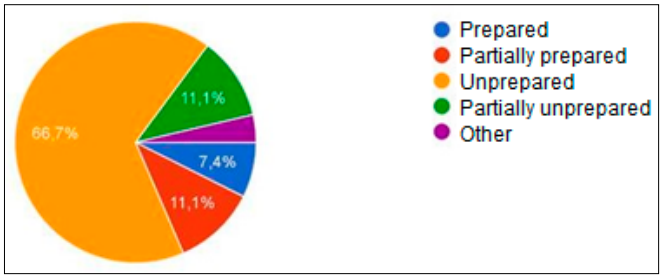
Graphic 2. Access to information about how to teach children with genetic syndromes while at education school. Source: Questionnaires answered from Aug. 5, 2020 to Oct. 9, 2020.

Only 14.8% of the participants reported that they had worked as educators with children with CCS (Graph 3).



Graphic 3. Experience working with children with CCS as an educator. Source: Questionnaires answered from Aug. 5, 2020 to Oct. 9, 2020.

When enquired about how well the teachers were prepared to deal with children with CCS, 66.7% felt unprepared and 7.4% prepared. Another 11.1% felt partially prepared and 11.1% felt partially unprepared (Graph 4).



Graphic 4. Preparedness in dealing with children with CCS. Source: Questionnaires answered from Aug. 5, 2020 to Oct. 9, 2020.

The four open-ended questions were answered only by the teachers with experience working with children with CCS. As indicated in Graph 3, only 14.8% of the teachers answered these questions, which probed into the obstacles teachers faced when teaching students with CCS. An interesting account reads as follows:

"The school gave me very little support when I had a child with CCS in my class. There was no one I could talk to. The only help I got was when I had to change the child's diapers and someone would walk in until I was back. I had to mix the food she would eat in a blender, since her mother said she might choke to death. As far as her education was concerned, I tried to adjust some of the tasks so that she could do something within her possibilities. It was a very complicated and challenging year."

This is what another teacher wrote about it:

"The first obstacle was the sheer number of students in class. A smaller class, possibly not as noisy and hectic, would have allowed me to do a better job. The second was to understand how the syndrome affected the student's learning. I was given some guidance by a supposedly qualified person, but it was always too generic. From the sixth grade on, the student starts to have contact with specialist teachers, differently from the reality of the early years of elementary school. Many times, adapting/adjusting content was exhausting, because there was no guidance to be followed."

In relation to teaching practices, the teacher cited above wrote that he often used images (pictures in a textbook, representations in the workbook, among others) and music. And he added: "Of course, I could not do it every day, but I did it whenever it was possible."

Unfortunately, many preferred not to answer the open-ended questions, which made it impossible to process them and provide more information.

DISCUSSION

The inquiries into CCS and the review of the literature performed as a part of this article indicated that few studies have been conducted about the development of children with the syndrome, especially in a school setting. As the questionnaire indicated, few teachers knew about CCS (14.1%) or had worked with children with the condition. Since this is a condition without a cure and with patient-specific outcomes, Drago et. al (2014) believe that children with CCS should be followed individually by a group of professionals acquainted with the syndrome and the limitations it imposes.

We also found that support and knowledge about genetic syndromes is lacking in schools, since most participants felt they were unprepared (66.7%) to deal with children with CCS.

Despite the progress seen in Brazilian legislation in matters related to inclusion, obstacles, prejudice, and unpreparedness are still the rule[6]. One cannot help but think that in order to experience inclusion, it is not enough to have favorable legislation; changes must be implemented in the education system to consider the wider socioeconomic context and the availability of specialist teachers in a setting of specialized educational care[3].

One of the respondents mentioned lack of support from the school and difficulty adapting content due to lack of knowledge. Another described the difficulties of managing a class with one or more students with special needs without a teaching assistant.

Our findings shed light on the lack of knowledge about CCS and on the role of families; when families do not understand their role in the inclusion of their children in school or if they lack support from the school throughout the process, the development of children with CCS is compromised[3].

Only two respondents cited the use of teaching resources to address the needs of students with CCS, reinforcing the difficulty and lack of support to do so, which often caused them to adapt class tasks whenever possible.

Education experts in general agree that there is much to be done for inclusive education to materialize in a satisfactory manner with quality and equity, in a way to take into account the specificities of each child with special needs without letting go of affection, commitment, and professional training[2,6,15,16].

As indicated in our survey and literature on the subject, educators possess little knowledge about CCS.

CONCLUSION

Our article offers reflections on CCS, its characteristics, and the difficulties associated with the inclusion of children with the syndrome in schools. In this endeavor, we gathered information about the disease and the inclusion of children with CCS in regular schools.

However, there was little literature on the subject, which prompted us to develop a questionnaire for elementary education teachers to find out more about the subject.

It is important to stress that much progress has been made, including in Brazilian legislation, in relation to inclusive education, although many obstacles and a lack of specialist teachers are still present, as indicated by questionnaire respondents.

We also found that the inclusion of children with CCS and other conditions must be substantially strengthened for inclusion to occur entirely.

In conclusion, we found that a very small number of teachers knew about CCS, its characteristics and difficulties, and that most had no contact with the disease in the classroom or during training. The path leading to the inclusion of children with CCS needs to be developed based on knowledge, affection, love, and respect for differences.

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ANNEX 1 - DIGITAL QUESTIONNAIRE - GOOGLE FORMS

RESEARCH – Cri Du Chat Syndrome

This form was designed to collect information for a Term Paper. Participation is voluntary and respondents are anonymized. The data collected in this questionnaire will be used only for scientific purposes.

*Required

1) Are you aware of the difficulties that Cri Du Chat Syndrome may impose on the development of children with the disease?*

Check only one box.

[ ] Yes, I am.
[ ] I am not.
[ ] I am partially aware.

2) During your training in the school of education, did you have access to information on how to work with children with genetic syndromes (such as Down syndrome, Edwards' syndrome, Klinefelter syndrome, Cri Du Chat Syndrome)?*

Check only one box.

[ ] Yes
[ ] No
[ ] Some

3) As an educator, have you ever worked with children with Cri Du Chat Syndrome?*

Check only one box.

[ ] Yes
[ ] No

4) How would you rate yourself in terms of your ability to deal with children with Cri Du Chat Syndrome?

Check only one box.

[ ] Prepared
[ ] Partially prepared
[ ] Unprepared
[ ] Partially unprepared
[ ] Other:

5) In case you have worked with children with Cri Du Chat Syndrome, what was the greatest difficulty you have encountered in the teaching-learning process?

_____

_____

6) Which pedagogical practices that you have adopted with children with Cri Du Chat Syndrome were the most successful in the classroom?

_____

_____

7) In your opinion, what is the role of educators in the development of children with Cri Du Chat Syndrome?

_____

_____

8) During your pedagogical practices with children with Cri Du Chat Syndrome, did you need resources that were not available at school?

Check only one box.

[ ] Yes
[ ] No
[ ] Sometimes