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Implications in Educational, Linguistic, and Psycho-Emotional Development in People with 22q11 Syndrome

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Abstract

22q11 Syndrome is one of the so-called rare diseases. This syndrome is characterized by certain physical alterations. However, its manifestations also occur in other areas. At the linguistic level, there is mainly an alteration in the production of language. In the educational area, the manifestations of the syndrome cause learning difficulties that will also vary depending on the cognitive level. All these characteristics, of course, have repercussions at the psycho-emotional level, with a greater tendency to the presence of mental disorders. This article aims to highlight which are the most important characteristics of this type of population and propose possible suggestions for intervention in order to minimize these problems. In addition, a series of recommendations are included to improve intervention at all levels and to improve the quality of life of people with 22q11 Syndrome.

Keywords: 22q11 Syndrome, education, language, psycho-emotional development

1. Introduction

The 22q11 syndrome is a genetic alteration caused by the loss of a fragment in region 11 of the long arm of chromosome 22, which causes a lack of genetic material (Fullman & Boyer, 2012). The incidence of this syndrome, also called DiGeorge syndrome, Velo-Cardio-Facial, or isolated cardiac abnormalities; It is around 1:2000-4000 (López-Rivera et al., 2017). Although it is manifestation is diverse, there is a physical pattern that is shared in all cases. They present the following characteristics: elongated face, narrow palpebral fissures, lowered corner of the lips, prominent nose, hypoplasia of the nostrils, dysmorphic ears, thin upper lip, and are oral breathers (Álvarez et al., 2009).

There are also alterations that develop in the majority of cases that present the syndrome, such as cleft palate or cardiovascular alterations (Young et al., 1980). A minority, around 10-20%, have a low blood calcium concentration or immunological disorders (Shprintzen et al., 1981; Junker & Driscoll, 1995). This causes alterations to appear due to structural and physiological anomalies, which causes specific alterations in speech such as articulatory difficulties in fricatives, affricates, trills, and some consonant groups (Sebastián-Lázaro et al., 2020).

2. Linguistic Characteristics in People with 22q11.

Regarding the linguistic characteristics, the language in people with 22q11 appears altered, due to the delay in the development of speech and language characteristic of this syndrome (Gerdes et al., 1999). Among these characteristics, a greater deficit in receptive language than in expressive language stands out (Glaser et al., 2002). In addition, delays appear, both in comprehension and in the use of vocabulary and syntax. In the area of semantics, alterations appear in the lexicon, manifested as a difficulty in memorizing new vocabulary and recalling it, as well as a low level of knowledge of the lexicon (Sebastián-Lázaro et al., 2020). Difficulties in pragmatics are the most characteristic of this disorder, including alterations such as difficulty adapting to the context, to the coherence or the comprehension of the interlocutor. In addition, the construction in the speech and the maintenance of the conversation turns are a difficulty in the case of 22q11.

In particular, disturbances in theory of mind are characteristic (Niklasson et al., 2002). They show difficulties interpreting humor and non-verbal communication, being very literal. However, some authors associate these alterations in theory of mind with verbal comprehension difficulties, since the evaluations have large verbal implications (Campbell et al., 2015).

Social communication is altered, due to the pragmatic profile presented by the subjects. This profile intervenes in social relationships, making them complex (Culler-Landsman, 2007; Niklasson et al., 2009). In children between 11 and 18 years old, it has been observed that they do not use abstract language, in addition, they have a misinterpretation of facial expression (Murphy, 2004; Van den Heuvel et al., 2017). It is worth noting studies such as those by Campbell et al., (2015) in which they show how adolescents have great difficulties in expressing and recognizing emotions.

3. Educational Characteristics in People with 22q11

Although a sizeable percentage of people diagnosed with 22q11 attend ordinary education schools (García & Pozo, 2021; Kok & Solman, 1995), we know that there are difficulties at the educational level that accompany children and adolescents throughout this stage (Swillen, 2001). It is interesting to note that, although part of this population has a diagnosis of moderate intellectual disability or borderline intelligence, learning difficulties appear even in cases with an IQ within the norm (Oliveira et al., 2018).

In general terms, the main difficulties are found in the mathematical area (Kok & Solman, 1995; Moss et al., 1999; Oliveira et al., 2018) and in reading comprehension (García & Pozo, 2021; Swillen, 2001). The origin of these problems may be due, in part, to Working Memory, specifically to visuospatial abilities; attention problems and problem solving (García & Pozo, 2021; Moss et al.,1999; Swillen, 2001).

Likewise, some strong points can also be highlighted, which are important to bear in this regard. For example, children with this syndrome tend to have facilities in learning through repetitive verbal memory and imitation of what they see around (Swillen, 2001). They also have excellent rhythm and musical sense, which allows them to excel in subjects such as music (García & Pozo, 2021).

It is essential to make visible the educational characteristics of these children so that both teachers and family members understand and see the limitations and strengths that allow improving the student's academic experience. Some studies have shown that students perform two or more years below their peers (Kok & Solman, 1995).

4. Psychoemotional Characteristics in People with 22q11

Overt language disturbances can contribute to difficulties in the school environment, leading directly to an increased risk of social and emotional problems and, in the long term, psychiatric disorders (Del Dotto et al., 1991).

There is increasing scientific evidence of the association of 22q11.2 Syndrome with psychopathology. Scientific evidence has shown over the years that there is a high prevalence of psychiatric disorders associated with 22q11 syndrome, although the figures vary depending on the studies carried out. For example, some authors establish the existence in 93% of the patients evaluated of at least one psychiatric diagnosis (Gothelf, 2007). Other authors find around 75% of the occurrence of different mental disorders (Green et al., 2009; Jolin et al., 2009; Ousley et al., 2013; Sobin et al., 2005; Tang et al., 2014). Between 40-76% is associated with anxiety disorders, 9-35% with mood disorders, 12-68% with ADHD, 23-41% with schizophrenia, 9-43% oppositional defiant disorder and 14-50% autism spectrum disorders (Schneider et al., 2014). It therefore seems clear that there is a psychopathological association with different psychiatric syndromes (Alberdi et al., 2018).

Some research suggests that there is a specific behavioral/psychiatric phenotype in 22q11DS (Swillen & McDonald-McGinn, 2015). Relative to the categorical approach to psychiatric diagnoses, individuals with 22q11DS show higher prevalence rates of Autism Spectrum

Disorder (ASD), Attention Deficit/Hyperactivity Disorder (ADHD), Mood/Anxiety Disorders, and Psychotic Disorders compared to people with idiopathic disorders (Fiksinski et al., 2018). Added to this is the fact that psychotic and mood disorders increase significantly throughout development (Vorstman et al., 2015). For example, in the case of the presence of schizophrenia, the figures vary from 5% in the case of young people to 40% in the case of those over 25 years of age (Murphy et al., 1999). This also happens in terms of behavior problems. In longitudinal studies, individuals with 22q11 have been shown to score significantly higher in areas such as withdrawal, thinking problems, attention problems, aggressive behavior, internalizing problems, externalizing problems, and total problems (Briegel et al., 2008; Briegel & Andritschky, 2021; Wagner et al., 2017).

Therefore, age seems to be a determining factor in the appearance of certain psychosocial-emotional characteristics. In childhood and adolescence, learning disorders and conduct disorders are the most frequently described psychiatric problems associated with 22q11.2, resulting in many cases being the major causes of morbidity of the syndrome (Martínez et al., 2011; Shprintzen, 2005). However, in adulthood affective disorders and psychotic disorders predominate. Hooper et al., (2013) present fundamentally two factors as predictors of the existence of subsequent psychopathology: the presence of a low IQ and a high score in externalizing and social behavior problems.

In the case of behavior problems in childhood and adolescence, a higher prevalence of internalizing problems has been found (Jansen et al., 2007; Brieguel, Schneider & Schwab 2008) and more social, attention and thinking problems, especially in those with an IQ below 70 (Jansen et al., 2007). Additionally, Jansen et al., (2007) stated that patients with 22q11 have comparatively more behavioral problems than patients with other genetic syndromes with the same levels of intelligence quotient, which could be indicated as a possible psychological characteristic of this group. Schneider et al., (2014) points out that disruptive behavior disorders represent 11% of this syndrome, with oppositional defiant disorder being the most frequent, especially in males.

5. Intervention in the 22q11 Syndrome Throughout Its Development

When conducting a treatment for this affectation, it is important to consider a necessary multidisciplinary intervention in these patients, as well as the application of these practices throughout the life cycle, paying special attention to childhood (Álvarez & Puth, 2022).

Regarding the linguistic intervention and due to the effects, that characterize this Syndrome, the sessions with the speech therapy professional will be based on the use of visual aids, structuring of the contexts so that they are predictable and previously known, and reinforcement of communication. in its social relations (Sebastían-Lázaro, Brun-Gasca, & Fornieles-Deu, 2020). Another of the intervention proposals in this area is that of Montserrat Molina Vives (2007) who advocates the re-education of language, emphasizing the development of linguistic and thinking skills; as well as the ability to interact in society.

As far as psychological intervention is concerned, control of both cognitive functioning is required; as well as behavioural, social, and mental health throughout life. These children can

benefit from behavior modification treatments (such as applied behavior analysis) and social interventions (with supervised play groups, play-based therapy, pragmatic training, peer-based social skills...) (Fourneret & Da Fonseca, 2019).

And, finally, in the intervention in the school environment, coordination between external and internal school professionals is presented as fundamental for a good management of the treatment, as well as use of technology with specific software. Specific and comprehensive care should be offered due to the variability of the characteristics and heterogeneity within this group (Álvarez et al., 2009).

6. Considerations for Working with People with 22q11 Syndrome.

As stated in the educational guide prepared by the Spanish Association of 22q1 Syndrome and the Spanish Confederation of People with Physical and Organic Disabilities (2021), it is important to take into account a series of guidelines and considerations when working with people with S22q11:

• Establish routines that anticipate the structure and content of the activities in order to favor the autonomy of people with 22q11.

• Encourage the establishment of eye contact by making them aware that they are not establishing it or positioning something striking over the interlocutor's eyes.

• Promote the performance of tasks that require little attentional demand or, failing that, perform various tasks in short periods of time.

• Work on the ability to inhibit impulses through board games such as Color Line.

• Try to make behavioral rigidity more flexible by favoring new interests.

• Use simple sentences, not exceptionally long and with a vocabulary according to the person's needs.

• Use, as far as possible, manipulative, and visual material that helps to make tangible concepts that are not.

• Encourage the use of adapters that thicken the barrel of paints and pencils to reduce fine motor skills.

• Provide support guides that help conduct activities that require several steps.

• Accompany the learning of reading and writing with sensory reinforcement.

• Continue to reinforce phonological awareness skills as an important foundation for learning to read and write.

• Develop custom dictionaries to enrich vocabulary as a basis for reading comprehension.

• Assess their written expression based on their level of oral language.

• Support communication with Augmentative and Alternative Communication Systems (SAAC).

• Avoid using irony, idioms or set phrases and, in case of using them, support expressions with images.

• Work on social skills through role-playing to promote the relationship with their peers.

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