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CASE REPORT

Marriage and reproduction in a woman with Down syndrome[a]

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Abstract
Sex life of people with Down syndrome (DS) or other conditions associated with intellectual disability is still a taboo, with few reports in the literature. Advances in knowledge of causal and nosological aspects, including its social achievements, have led to the strengthening of the inclusive movement aimed at those people. This paper presents an unusual case of successful marriage and reproduction of a woman with DS. The proband studied in special schools and communicates well verbally. She presented menarche at age of 13, showing autonomy in caring for her body. Eight years ago she met her current husband at the special school she attended. Two years after the wedding, the proband became pregnant of a male child without the syndrome. She is able to take care of her child needs, sharing this responsibility with her own mother, who was primarily responsible for her education directed towards autonomy. The proband's karyotype revealed trisomy 21 with chromosomal mosaicism. New social achievements are occurring, among them the establishment of lasting emotional relationships. The reproductive chances and risks of recurrence of DS should be considered in genetic counseling. The breeding and rearing of any children born from these marriages become new responsibilities shared by these special parents and their families.

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PALABRAS CLAVE
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Discapacidad intelectual;
Matrimonio;
Reproducción

El matrimonio y la reproducción en la mujer con síndrome de Down

Resumen
La vida sexual de las personas con síndrome de Down (SD) u otros trastornos asociados con la discapacidad intelectual sigue siendo un tabú, con pocos relatos en la literatura. Los avances en el conocimiento de los aspectos causales y nosológicos, incluidas sus conquistas sociales, llevaron al fortalecimiento del movimiento inclusivo destinado a estas personas. En este artículo se presenta un caso inusual de un matrimonio de una mujer con SD que tiene un hijo. Esta mujer estudió en escuelas especiales y se comunica bien verbalmente. Presentó menarquía a los 13 años de edad, y demostró autonomía en el cuidado de su cuerpo. Ocho años atrás conoció a su actual esposo en una escuela especial. Después de 2 años de matrimonio, ella quedó embarazada de un varón sin este síndrome. La mujer es capaz de atender las necesidades de su hijo, responsabilidad que comparte con su madre, que fue la principal responsable de su educación hacia la autonomía. El cariotipo de la probanda reveló trisomía 21 con mosaicismo cromosómico. Se están produciendo nuevos logros sociales, incluido el establecimiento de relaciones afectivas perdurables. Las posibilidades de reproducción y el riesgo de recurrencia del SD deben ser considerados en el consejo genético. El cuidado de todos los niños nacidos de estos matrimonios genera responsabilidades compartidas por estos padres especiales y sus familias.

Introduction

Down syndrome (DS) is an archetype of congenital genetic disorder. Advances in knowledge of causal and nosological aspects, including its social achievements, have led to the strengthening of inclusive movement aimed at people with intellectual disability and congenital disorders in general.

This syndrome was first described by the English physician John Langdon Down (1866)[1], but its etiology was clarified only in 1959, when the geneticist Jerome Lejeune and his collaborators associated this syndrome with the presence of an extra chromosome 21, then becoming known as Trisomy 21. This was followed by other studies on the pathogenesis of this chromosomopathy characterizing it as a free trisomy, by translocation or mosaicism, when normal and trisomic cells are present, which occurs in 1-2% of the cases[2-4]. The free trisomy 21 constitutes the most common form of DS and has been associated with an increase on its rates when considering maternal age above 35 years[5].

Down syndrome is characterized by mild to moderate intellectual disability, hypotonia and typical craniofacial signs such as oblique palpebral fissures. Heart disease and other severe systemic disorders occur in lower percentage of cases. The trisomy of 21q22 chromosomal band is considered pathognomonic and related with more severe disorders, although there is considerable phenotypic variation[6]. In cases of mosaicism, there are clinical manifestations ranging from a typical presentation of DS signals even very subtle, often only detected after birth[7]. The more severe symptoms may be associated with a higher percentage of trisomic cells[8] or with loss of chromosome 21 in a zygote originally with trisomy.

Social inclusion and education has also contributed to the development and adaptation of people with DS, often leading to possibilities of working activities in conditions adapted to their characteristics[9]. Nevertheless, the sex life of people with this syndrome or other conditions associated with intellectual disability is still a taboo, with few reports in the literature. The objective of this paper is to review different aspects related to emotional and sexual development in DS reported through the life story of a DS woman with lasting marital relationship that led to the birth of a child without the syndrome, emphasizing the importance of the family in this process.

This paper presents an unusual case of successful marriage and reproduction of a woman with DS attended at the Program of Community Genetics (Genetics & Society), Federal University of Bahia, Brazil.

Clinical observation

The study refers to a women with DS, now with 41 years old. Family history is negative for congenital malformations and genetic disorders. Mother aged 20 and father aged 25, in her birth. She was born of vaginal section without complications. Clinical diagnosis of DS, in absence of severe congenital malformations. Later, she had two siblings. She studied in special schools; made no speech therapy, but can communicate well verbally. She had swimming lessons and dance. The father is a musician and publicist and she was always passionate about music. At home she was treated in the same way that her brothers without the syndrome were. She could not be literate, but was prepared for the job market. She worked in commerce and recently retired.

The proband presented menarche at age of 13, showing autonomy in caring for her body. She dated three colleagues, one with the same syndrome, but was looking for a companion for a family. Eight years ago she met her current husband at the special school she attended. He was a orphaned boy with a history of mild learning deficit associated with gestational events, which lived and worked in a specialized institution.

The wedding came naturally, as any couple (Figure 1 A). They married in a religious ceremony and came to live with the maternal family, dividing tasks and bringing some financial contribution to the home. Two years after the wedding, the proband became pregnant at the same time as her sister-in-law, surprising everyone, because the parents had received information that there was no need for contraceptive methods because, due to the syndrome, there was no chance of pregnancy. The prenatal examination, performed at 5 months of gestation, indicated that it was a male child without the syndrome, news received with great joy by the entire family.

After the birth of her son, the proband, that since her birth had received only clinical diagnosis of DS, conducted the study of her karyotype, which revealed trisomy 21 with chromosomal mosaicism. Karyotype: 47,XX,+21/46,XX, with the majority of the trisomic lineage, present in 80% of examined cells. Currently, she enjoys good general health. Participates in literacy course for adults and is engaged in educational activities about DS. She is able to take care of her child needs, sharing this responsibility with her own mother, who was primarily responsible for her education directed towards autonomy. In six years of marriage, the couple maintains a stable and loving relationship, dedicating their child to leisure time (Figure 1 B).

Discussion

A historical analysis of the development of people with DS shows a change of conceptions about this syndrome, initially based on superstitions and prejudices, followed by a pathological conception of the disease; and currently considered due to a genetic alteration compatible with life and possible adaptation in society as a part of human diversity, with rights and social duties. It sets up a new interpretation model for disability, in which it loses the character of individual attribute and is now considered a contingent phenomenon that reshapes social actions and adjusts the environment regarding the nature of people with disabilities[10].

Advances in life's quality, longevity and inclusion measures has enabled new social roles, however the issue of sexuality has been ignored or underestimated. Brown (1996)[11] observes that relationships and marriages are in the context of quality of life in DS and calls attention to the need to prepare these people for life, so that these possibilities can happen. Denholm (1992)[12] refer that adolescents with intellectual disabilities have the same expectations in terms of moral codes, friendships, interests in fashion that young people without disabilities and argues that these trends may become more widespread, though they have less social opportunities. Conod and Servais (2008)[13] report a lack of studies on people with intellectual disabilities in terms of sex as well as relationships, marriage and parenting, also remembering that these activities do not only depend on the people's expectations, but also of socialization opportunities offered.

The families, especially the parents, are expected to be better prepared to attend the needs of their children in building their personality and evolutionary changes in various aspects of social life, and there must be no difference regarding to the DS's ones. Even with all the independence that a person with DS can achieve, there is always a consideration, support, in which the family is requested with greater or lesser frequency[14].

Today, depending on the case, the sexuality is more accepted by the Down people relatives, but the issue of reproduction is viewed with caution and seen as unfeasible by 70% of parents[15]. In men with DS, despite the normal development of secondary sexual characteristics, fertility is reduced, possibly due to the anomalous behavior of chromosome 21 in male meiosis[16]. Goldstein (1988)[17] refer that in women with DS, sexual development occurs in a manner similar to people in the general population in terms of pu-

Figure 1 Couple in the beginning of the relationship (A) and gathering the family (B).

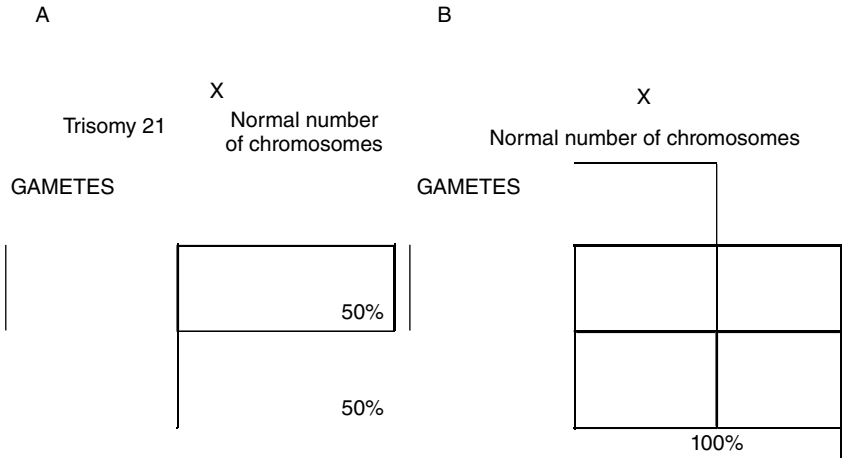

Figure 2 Segregation of chromosome 21 and probabilities of trisomic (A) and normal (B) zygotes in reproducing woman with chromosome mosaicism.

berty and sexual maturation, with an average age of menarche at 13.6 years, compared to 13.5 in controls.

Reviewed data on pregnancies of 26 DS women with no mosaicism, shows ten SD children, 18 without chromosomal disorders and 3 miscarriages[18]. According to Moreira and Gusmão (2002)[19], when the couple is formed by another person without the syndrome, the genetic risk of SD progeny is about 50%. In SD mosaic, such as presently reported, the possibilities of fetuses with DS are lower (Figure 2) and depend on the proportion of trisomic cells on the gonadal tissue.

In this case, the fundamental premise of the proband's parents was to develop her autonomy, with clarifications and specific support for their uniqueness as well as the belief in their possibilities of development and realization of their dreams. This same attitude remains currently in the grandson's mediation towards education and support to the couple.

The case presented here demonstrates that in an inclusive environment, social opportunities can occur and issues such as marriage and reproduction in DS should be evaluated in the context of the family.

Conflict of Interest

The authors declare that they have no conflict of interest.

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