EMOTIONAL AND COGNITIVE ASPECTS AND QUALITY OF LIFE IN ADOLESCENTS WITH DISORDERS OF SEX DEVELOPMENT: CASE STUDIES

Aspectos emocionais, cognitivos e qualidade de vida em adolescentes com desordens do desenvolvimento sexual: estudos de caso

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ABSTRACT

This study describes emotional, cognitive and quality of life aspects of adolescents diagnosed with Disorders of Sex Development (DSD). It is a multiple case study that analyzed medical and psychological longitudinal records of 5 adolescents with DSD and female sex of rearing. The participants also performed a battery of emotional and cognitive tests. Similarities were found among the 5 participants, consistent with the adolescence stage and their conflicts. Body image concerns and desire for the development of secundary sexual characters were identified among them, as well as the maintenance of infantile behaviors as a way to avoid dealing with sexuality. Avoidance strategies are used to deal with anxiety arising from the process of transition into adult life.

Keywords: Disorders of sex development (DSD). Adolescence. Emotional evaluation. Developmental psychology.

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RESUMO

O estudo descreve o desenvolvimento emocional, cognitivo e a qualidade de vida em adolescentes com diagnóstico de Desordens do Desenvolvimento Sexual (DDS). Foi utilizado delineamento de estudos de casos múltiplos. Foram analisadas as fichas de acompanhamento psicológico longitudinal de 5 adolescentes com DDS com sexo de criação feminino, que também foram submetidas a uma bateria de testes para avaliar aspectos emocionais e cognitivos. Semelhanças foram encontradas entre as 5 participantes, condizentes com a etapa da adolescência e seus conflitos. A preocupação com a imagem corporal e o desejo pelo desenvolvimento de caracteres sexuais foram identificados entre as mesmas, assim como a manutenção de comportamentos infantis como forma de defesa contra a sexualidade. Estratégias evitativas são utilizadas para lidar com a ansiedade decorrente do processo de transição para a vida adulta.

Palavras chave: Desordens do desenvolvimento sexual (DDS). Adolescência. Avaliação emocional. Psicologia do desenvolvimento.

1 INTRODUCTION

Disorders of Sexual Development (DSD) involve a large group of conditions caused by failures during sexual determination and/or sex differentiation in individuals, during the women's pregnancy (Cools, Nordenström, Robeva, Hall, Westerveld, Flück, et al., 2018). The diagnosis of DSD encompasses a wide variety of conditions and etiologies that should be studied and understood separately (Schweizer, Brunner, Gedrose, Handford & Richter-Appelt, 2017). The most complex clinical manifestation is genital ambiguity. These patients require integrated care of several medical specialties in addition to psychological counseling, preferably in life-long follow-up mode (Lee et al., 2016).

Adolescents with DSD become aware of their sexual development diferences when facing the expectation of puberty and its body changes. The diagnosis of DSD can devastate adolescents' identity and self-esteem, as well as impact their family (Lee et al., 2016). Psychological stress, risk behavior, and suicidal tendencies were identified in higher frequency and intensity in these patients, compared to the general population (Schützmann, Brinkmann, Schacht & Richter-Appelt, 2009).

Although this subject is widely discussed in the literature, there are still a lack of knowledge on the psychological development, especially during adolescence (Crouch & Creighton, 2014; Kleinemeier, Jürgensen, Lux, Widenka & Thyen, 2010; Schweizer et al., 2017; Thyen et al., 2018). This study describes five case studies of adolescent patients with DSD, considering cognitive and emotional aspects as well as perception of quality of life.

2 METHOD

This study belongs to a longitudinal research aimed at evaluate the psychological, cognitive and quality of life aspects in patients with DSD. Participants are patients from the DSD Multidisciplinary team (PADS) at Hospital de Clínicas, Porto Alegre. The PADS has attended 156 patients since its inception. Of those, 11 individuals were between 14 and 16 years old in 2018. All of them were invited to participate. However, 3 had lost follow-up and 3 refused to participate in this study. Five participants then composed the sample. It is a qualitative and exploratory-descriptive study, presented in multiple cases format, as proposed by Yin (2004).

Ethical aspects were assured through resolution number 466, of December 12th 2012, from Conselho Nacional de Saúde. The Informed Consent were read to and signed by the patients and/or their parents or legal representative. The research was approved at Ethical Committee of Hospital de Clínicas de Porto Alegre: CAAE number 79027717800005327.

Participants

Five adolescents diagnosed with DSD, assigned as female at birth, aged 14 to 16 years, receiving treatment at PADS. Characteristics such as diagnosis, karyotype, sex of rearing and age of diagnosis are described in Table 1.

Table 1

	CASE 1	CASE 2	CASE 3	CASE 4	CASE 5
Age (years)	14	14	15	16	16
Diagnosis	WT1 mutation	SW CAH	SW CAH	CAIS	SW CAH
Karyotype	46,XX	46,XX	46,XX	46,XY	46,XX
Sex assignment	F	F	F	F	F
Age of Diagnosis	At birth	During pregnancy	At birth	3 yrs old	At birth

SW CAH: salt-wasting Congenital Adrenal Hyperplasia

CAIS: Complete Androgen Insensitivity Syndrome

Procedures

Data were obtained from both psychological records and evaluation. These records were analyzed retrospectively, considering data from anamnesis, diagnosis of DSD and its treatment, life history, and common emotional aspects observed.

The tests were divided into two steps to avoid the fatigue of the participants, with a time interval of a couple of months. In the first stage, patients performed Wechsler Abrevieted Intelligence Scale (WASI), House-Tree-Person Test (HTP) and The World Health Organization Quality of Life Assessment (WHOQOL-bref). In the second meeting, Rorschach and the Beck Depression Inventory (BDI) were applied. All tests were conducted according to the instrument's manual.

Data analysis was performed according to Yin (2004), who proposes three phases: (1) choosing a theoretical framework for understanding the

phenomenon (Psychoanalysis), selecting cases and using instruments; (2) data collection, with exhaustive reading of the descriptive material and survey of each patient's tests; and (3) identification of the themes related to the study objective, identifying both singularities and similarities among the participants. Each participant represents a unit of analysis, allowing to emerge its unique aspects, as well as similarities among them.

3 RESULTS

In order to preserve anonymity of the participants, they are named with case number in parentheses (i.e.: CASE 1). The other family members will be called with the initial of the noun, associated with the case number (example: M1, for mother of participant 1).

Case 1 – 14 years old

M1 underwent an emotional breakdown in the first months of the CASE1´s life, losing clinical follow-up at the hospital of origin. The patient was referred to PADS around six months old, with virilized genitalia. Within one year of age, vaginoplasty and clitoroplasty were performed, along with gonadal biopsy. The anatomopathological result was rudimentary testis.

CASE 1 is the second child of the couple. M1 experienced a troubled relationship with F1. He was addicted to alcohol and unemployed. The couple did not plan the pregnancy and did not do layette. M1 had high expectations of having a boy. No ultrasound was performed during pregnancy. The mother had a hard time accepting a girl. A friend, CASE 1's godmother, who latter became CASE 1's stepmother, helped her. However, she kept with emotional instability.

CASE 1 had a regular development during childhood, overcoming the evolutionary tasks. Her parents divorced when she was two years old and her mother had multiple conjugal relationships. M1 says she had always thought the daughter would grow up to be homosexual, since a lifelong observation of preference for toys such as cars, balls and sports games.

Only at eight years old, CASE 1 did psychological monitoring for a few months, due to conflicts generated by low self-esteem and school difficulties. At

11 years old, when her mother definitely left her, she resumed psychotherapy. She was living with her father and stepmother. CASE1 expressed family conflicts, as well as the implications of the diagnosis in her development.

Her molecular diagnosis resulted in a DSD due to pathogenic variation in the WT1 gene. Currently, CASE 1 understands her diagnosis; however, she is insecure and has questions regarding her sexuality and gender identity. CASE1 has had only one romantic relationship, which was heterosexual. She has not had sexarche and has a prepubescent appearance. Although she is 14 years old, she does not want to develop female sexual characters: She is afraid of having a woman's body, preferring to maintain her childish appearance. Her family is available and adherent to the proposed treatment, as well as able to maintain an effective dialogue with the patient and care team.

The cognitive assessment, based on WASI, indicates that the general cognitive ability of CASE 1 was classified as Borderline, with Intelligence Quotient (IQ) of 79. She presented a significant difference between the verbal area (QIV = 97) and the execution area (QIE = 67). It indicates issues in functional life, with difficulty in applying her intelligence. The HTP test indicates feelings of insecurity and withdrawal attitude when facing social interactions. She presented difficulties in the sexual area, with her identity still in development.

In the Rorschach projective test, CASE 1 was able to adapt to the task and produce appropriately. She demonstrated a child's vision of reality, using escape and fantasy, all aspects usually observed in adolescents. She presented diffuse anxiety associated with a low tolerance. She tends to react with intense emotional components in the behavior when facing social interactions. She is in the process of developing her capacity to contain instinctive and impulsive reactions while maintaining authenticity. Her relationships are fearful, cautious and controlling.

The result in BDI corresponds to a minimum level for symptoms of depression. In the WHOQOL-bref, she presented results considered regular in the Physical, Social Relations and Environment domains. However, she was dissatisfied with the Psychological domain.

Case 2 – 14 years old

DSD was identified from an ultrasound, suspected of Congenital Adrenal Hyperplasia (CAH). M2 was referred to a tertiary service for prenatal care. The diagnosis was confirmed after 58 days of investigation. She had an episode of hydro electrolytic disturbance during this period, while hospitalized in Neonatology Intensive Care Unit. She received a female sex assignment and underwent vaginoplasty and clitoroplasty surgery at seven months of age.

M2 and F2 began their relationship during adolescence. They got married and planned for ten years to get pregnant. Couple's relationship has always been stable, and they had expectations of having a girl. They had already chosen the name even before the pregnancy and M2 identifies that, even before birth, the daughter already had an identity in her parents' imagination. When they heard of the possibility of a DSD, M2 said she was immensely shocked. The contact with the assistance care professionals and the information received helped her to understand the condition. After the diagnosis, the parents waited anxiously for surgery to correct the genitalia, reporting feeling relieved after it.

CASE 2 had a regular development during childhood, overcoming the evolutionary tasks. Family members were able to support the parents to deal with the condition and its treatment, since the condition was not kept secret. CASE 2 was considered a moody, bitter child and, at times, opposite. At school, she presented some difficulties that were attributed to hyperactivity and impaired attention. CASE2 has always preferred physical education. Until shortly before puberty, M2 was concerned with the disclosure of the diagnosis to the patient. She wanted to tell her daughter about her condition, but she did not know how to do it. With the support from the multidisciplinary team, she was able to communicate the diagnosis to her daughter.

Currently, CASE 2 is an extroverted and communicative teenager. She talks with the mother about their medications, treatment, and surgeries. She recently had psychological counseling for anxiety caused by a new surgical indication. CASE 2 considers herself a girl, wants her female sexual characters to develop and is satisfied with her appearance. She has not had either menarche or sexarche. She considers herself heterosexual, although she has

not had sexual experiences. Nowadays, CASE 2 knows about her diagnosis and administers her medications, demonstrating autonomy. She has medical indication to vaginal dilation, but postpone it justifying there is a long way before having an active sexual life. She has an adequate school performance. They have proper family support. The family is participative and adherent to the treatment proposed by the team.

Cognitive assessment, based on the WASI, indicates that the general cognitive ability of CASE 2 was adequate to age, classified as Medium (QIT = 90). There was no significant difference between the verbal area (QIV = 87) and the execution area (QIE = 93). The HTP test indicates insecurity and anxiety, with a withdrawal attitude.

In the Rorschach projective test, CASE 2 was able to adapt and produce appropriately. She demonstrated a child's vision of reality, using escape and fantasy, aspect commonly observed in adolescents. CASE 2 demonstrated traits of impulsivity, with difficulty in managing her emotional needs, an aspect expected in adolescence. However, she has insight and frustration tolerance.

The result in BDI corresponds to a minimum level for symptoms of depression. In WHOQOL-bref, she presented results considered regular in the Physical, Psychological and Environmental domains. However, she showed dissatisfaction with the domain of Social Relations.

Case 3 – 15 years old

The neonatologist diagnosed CASE 3 shortly after delivery due to undifferentiated genitalia. After 20 days of investigation, with the results of laboratory and imaging tests, Classical Congenital Adrenal Hyperplasia (CAH) was diagnosed. She received a female sexual assignment, and underwent vaginoplasty and clitoroplasty during her first year of life.

M3 and F3 have known each other since the end of adolescence. M3 became pregnant soon after marriage. The couple's relationship was stable. M3 wanted a boy and F3 a girl. When the child was born and diagnosed with DSD, the mother was in shock. She did not understand the diagnosis and felt distressed during the period of the investigation, in which the baby remained without a sex definition and civil registration. She had made a pink baby

trousseau because the ultrasound indicated a female baby. The contact with the assistance care professionals and the information received helped her in understanding the condition. After the sex assignment, the parents waited anxiously for surgery to correct the genitalia.

CASE 3 had a healthy development in childhood, managing to overcome evolutionary tasks appropriately. At three years old, she was referred for psychological evaluation due to a maternal complaint of aggression. At this time, the patient had, clinically, genital virilization. M3 also worried about her daughter's preference for stereotypically boys´ toys, fantasizing that she would be homosexual in the future. The patient kept psychological counseling for approximately two years aimed at working with age conflicts and at understanding her clinical condition. Currently, CASE 3 behaves like a quiet teenager. She has good school performance and likes sports.

With a female appearance, CASE 3 had menarche, but not sexarche. She has had heterosexual relationships. Throughout psychotherapeutic follow-up performed at age 13, she expressed a desire to be more feminine and to have sexual characters developed. Currently, she is more mature and aware of her health condition.

The family looks participative, but demands CASE 3 to have autonomy in administering her medication. However, she shows little adherence. She has medical indication of vaginal dilation, but refuses the procedure, justifying that she does not want to start a sexual life yet. Cognitive assessment, based on the WASI, indicates that her general cognitive ability was adequate to age, classified as Medium (TIQ = 98). There was no significant difference between the verbal area (VIQ = 92) and the execution area (EIQ = 103). HTP test indicates the need for support and defensive and withdrawal attitudes.

In the Rorschach projective test, CASE 3 was able to adapt to the task. She demonstrated a child's vision of reality, using escape and fantasy, aspects usually observed in adolescents. When facing pressure, CASE 3 became introspective. With insight and self-criticism, she is developing the ability to deal with suffering and maintaining authenticity and spontaneity. However, she has intense emotional components in her behavior, which impacts relationships.

She deals with it trying to avoid her feelings, developing fearful and cautious relationships.

The result in BDI corresponds to a mild level for symptoms of depression. In WHOQOL-bref, she presented results considered regular in the Physical and Environmental domains. However, she was dissatisfied in the areas of Psychology and Social Relations.

Case 4 – 16 years old

CASE 4 had no diagnosis of DSD at birth since she had female external genitalia. At three years of age, she was referred for inguinal hernia repair. During the procedure, gonadal structures were identified Anatomopathological and karyotype results´ exams were of testicles and 46, XY karyotype, confirming the diagnosis of Complete Androgen Insensitivity Syndrome (CAIS). Sex assignment had already been female.

CASE 4 is the third child of M4 and F4 who already had two male children. Her mother reports wishing very hard for a baby girl and, when the kid was diagnosed with CAIS, the mother needed psychological counseling. She expressed fantasies regarding the child's sexuality and gender.

CASE 4 had a regular development during childhood, overcoming the evolutionary milestones. She is described as a quiet and curious child, who enjoys studying and getting involved with scientific activities at school. She prefers activities such as reading, scientific projects and watching TV series and films. Currently, CASE 4 is satisfied with her appearance, despite expressing the desire to have bigger breasts. She is convinced of her female gender identity. She found out about her diagnosis at the age of 14 and sought confirmation with her mother. She is aware of her clinical condition and has received psychological counseling at various times in her life. Recently, she received psychological counseling to deal with typical changes of transition to adolescence. The patient and her family are participative and adherent to the treatment proposed by the healthcare team. She has an indication of performing vaginal dilation, but refuses it at the time, claiming she has no interest in starting a sex life.

Cognitive assessment, based on the WASI, indicates that her general cognitive ability was classified in the Lower Middle level (TIQ = 87). There was no significant difference between the verbal area (VIQ = 87) and the execution area (EIQ = 92). The HTP test indicates characteristics of insecurity and withdrawal attitude, associated with concerns about the surroundings and future.

In the Rorschach projective test, CASE 4 presented the ability to adapt to the task, despite some resistance. She demonstrated capacity for synthesis and abstraction, despite a relative impairment in the capacity of objective perception of reality, an aspect observed in this age group. When facing situations of pressure, she resorts to introspection and, through the use of self-criticism, seeks to elaborate her suffering, maintaining authenticity and spontaneity. She may present more intense emotional components in behavior, which is expected in adolescence.

The results in BDI correspond to a mild level for symptoms of depression. In the WHOQO-bref, she presented results considered to be regular in all domains.

Case 5 – 16 years old

After verification of undifferentiated genitalia (CASE 5) at birth, she stayed for two months in the hospital where she was born waiting etiological diagnosis and the sex assignment. From the laboratory and imaging tests, a Classical Congenital Adrenal Hyperplasia (CAH) was diagnosed, being designated female sex of rearing. CASE 5 underwent vaginoplasty and clitoroplasty surgery at two years old. She was referred to the center where this work is being performed at age 15, after losing follow-up at the hospital of origin. Since she has been in this center for a year, data on anamnesis and early childhood are lacking.

M5 reports that pregnancy was not planned and that her relationship with F5 was casual. The father did not participate in the upbringing of CASE 5. M5 states that the diagnosis at birth and the impossibility of knowing the sex of the daughter brought intense emotional distress, becoming depressed at the time.

She received support from her mother. M5 reports that her daughter was a quiet and insecure child, with dependent behavior.

CASE 5 started specialized multidisciplinary DSD treatment at puberty. The family reports that they have had psychological counseling in two moments before the surgery in the other hospital.

CASE 5 can explain her clinical condition, but she does not fully understand treatment and prognosis medication to be used or about the prognosis, showing immaturity and affective blunting. She is satisfied with her looks, however desires bigger breasts. She is convinced of her female gender identity and wishes for a more feminine body. CASE 5 had menarche, but not sexarche. She has not had romantic relationships yet. The patient´s support system consists of her mother and her maternal grandmother, with whom she can talk about the clinical condition.

Cognitive assessment, based on the WASI, indicates that the general cognitive ability of CASE 5 was classified at the Border level (TIQ = 72), as well as verbal area (VIQ = 77) and execution area (EIQ = 74). The HTP test indicates insecurity, with withdrawal attitude and need for support.

In the Rorschach projective test, CASE 5 presented the ability to adapt to the task. She demonstrated a child's vision of reality, using escape and fantasy, aspects commonly observed in adolescents. With low frustration tolerance, she demonstrated lack of flexibility and spontaneity. She maintains an avoided and reserved attitude, which is reflected in a fearful and cautious relationship.

The result in BDI corresponds to a minimum level for symptoms of depression. In WHOQOL-bref, she presented satisfactory results in the Physical, Psychological and Environmental domains; and regularly in Social Relations domain.

4 DISCUSSION

The absence of the child´s sex designation poses a significant emotional distress on the parents (Hemesath, de Paula, Carvalho, Leite, Guaragna-Filho & Costa, 2019), as showed in the participant´s mothers' reports. The surgery for the genitalia adjustment at birth has been widely discussed in the scientific

milieu (Gardner & Sandberg, 2018) and is subject of substantial controversy. However, all the mothers of this study, except for M4, who had not had the child's diagnosis at birth, reiterate the expectation of the surgery as soon as possible, after the sexual designation. Surgery was perceived by mothers as having the potential to validate the child's gender, generating relief in the family and aiding in family and social integration, as supported by studies of Hemesath (2013) and Freda, Dicé, Auricchio, Salerno & Valerio (2015).

Surgical procedure, according to the mothers' perception, allowed them to psychically represent the child with a defined sexual identity. It incentivizes parents to adopt parental strategies consonant with the sex assignment. CASE 1, however, adopted a skeptical stance on the sexual identity of her child. Her ambivalence was expressed in her beliefs that her daughter would be homosexual or transsexual because of DSD. CASE 1 is the only one who has shown insecurities and questions regarding her gender identity and sexual orientation, expressing dysphoria regarding the development of female sexual characters. In the other hand, CASE 2, CASE 3, CASE 4, and CASE 5 are satisfied with their sex assignment, wishing their bodies to be more feminine.

Cohen-Kettenis et al. (2010) allert that male gender behavior in patients with DSD and female sex of rearing should not be confused with male gender identity. Berenbaum (2015) has shown that there is a preference for male toys in girls with CAH, corroborating the findings of the present study. The author attributes these preferences, to a certain extent, to the prenatal effects of androgens. M1, M2, M3 and M5 reported preference for activities involving physical mobility and sports and choice of characteristically male toys in childhood, which worried the parents at the time. Due to brain impregnation of testosterone, some school age and puberty girls with CAH present male behavior and homosexual. It is common for them to seek psychological counseling, since revealing these situations for the family might be stressful and anxious (Thyen et al. 2018).

Based on the cases presented, it can be seen that the psychosocial construction of adolescents begins before birth, engendering aninfinity of fantasies and fears in the parental couple that serves as the basis for their understanding of the child. The initial shock of the diagnosis described by M1,

M2, M3 and M5, along with the temporary undefinition of sexual identity of the baby and the fantasies connecting DSD and sexual orientation and gender identity, seems to provoke overprotective behaviors from parents. The kids, by their turn, seem to "learn" to maintain a withdrawn and avoidant behavior, which is expressed in the five participants of this study, as well as in the results of the projective techniques HTP and Rorschach.

The adolescent, from the point of view of psychoanalysis, is a individual in the process of transformation, inserted in a profound revision of his/her inner world and infantile inheritances, working on adapting to the new body and the impulses from puberty. In this phase, the individual goes through a review of previous representations, constructed during childhood, and engages in the development of new representations, considering the new body, relationship's objects and the world (Levy, 2013). Patton & Viner (2007) describe the developmental transitions as dynamic phases, which can be characterized by stress, disruption and discontinuity, generating challenges and risks, as well as new opportunities. Biological changes include brain development that leads to the growth of cognitive abilities and body changes, which in turn lead to secondary sexual characteristics. In the cases presented, although they expressed a desire to develop a more feminine body, it seems to be a barrier to the establishment of affective/romantic relationships of any nature, adopting an avoidant attitude. They delay or refuse the vaginal dilation procedure (CASE 2, CASE 3 and CASE 4) stating that they have no interest in having intercourse.

Crouch and Creighton (2014) call attention to the transition process of adolescents with DSD into adult life, showing ambivalent feelings. It might be difficult to leave pediatric care and move forward, and to develop autonomy required in the adult teams (gynecology, for example). Likewise, it is also difficult for these patients to give up the presence of the mother (or parents) in the appointments.

CASE 1, CASE 2 and CASE 5 maintain childlike functioning and extend the dependency relationship with their parents. CASE 4 adopts defensive behavior, making use of a repertoire of rationalization and intellectualization in order to deal with the demands of the transition to mature life. On one hand she seems to understand in detail her clinical condition and to feel comfortable with

it; however, on the other hand, she avoids vehemently potential affective relationships and the experience of sexuality. CASE 3 demonstrates conflicts expected for the adolescence period: she performs transgressions to the treatment, which is described in adolescents with this condition, especially in cases of CAH (Hamed, Metwalley & Farghaly, 2018; Kleinemeier, Jürgensen, Lux, Widenka & Thyen, 2010). Compared to the others, however, she was the one who advanced most in the experience of her sexuality. Her parents want her to have greater autonomy in her treatment, however, she resists by complaining about "being different" from her peers. As shown in the results of the projective tests, there is an emotional difficulty in the participants in the face of the changes that occur in the transition to adult life. Through the Rorschach technique, the participants presented an infantile vision of reality, making excessive use of fantasy and missing aspects of objective perception. Although common for adolescents; it was more intense than expected. The adolescents have shown low frustration tolerance and use of defensive mechanisms that leads to the development of fearful and cautious relationships. These defenses and relationship characteristics reflect the difficulty in dealing with the conflicts and themes of adolescence, especially concerning sexuality. Maturity, early sexual life, and appropriation of one's own body are postponed (Kleinemeier et al., 2010; Schweizer et al., 2017).

The results of the HTP test corroborate the literature, showing feelings of insecurity and withdrawal. Another point raised by the HTP test was an excessive preoccupation with future sexual life, which is related to withhold of dilation procedure or avoidance of thinking about initiating sexual life. They are involved in anticipatory anxiety related to romantic and sexual relationships, as evidenced by Lee et al. (2016). Kleinemeier et al. (2010) elucidate that adolescents with DSD with female sex assignment are less sexually active. One of the hypotheses for such a phenomenon is that the inhibition may be caused by uncertainty about body anatomy and anxiety in talking to a potential partner about their diagnosis.

Regarding cognitive evaluation, two of the participants (CASE 1 and CASE 5) showed a significant reduction in the Intelligence Quotient (IQ). It is known that intelligence is one of the facets of the personality, being influenced

by the context and relations, as well as by some organic diseases (Brito, Schaab & Remor, 2018). Analyzing the particularities of each case, the borderline IQ of CASE 1 can be caused both by the troubled emotional context that she experienced during her development and by the hormonal deficit caused by her condition (WT1 mutation). CASE 5 had little adherence to treatment for many years of childhood, which has had an influence in the development of cognitive abilities, as shown in patients with CAH (Hamed, Metwalley & Farghaly, 2018).

As analyzed by Schweizer et al. (2017), one protective factor against psychological distress is to rely on family members and assistant care for support. The possibility of obtaining information and knowledge about diagnosis and etiology of DSD throughout their lives was pointed out as for developing coping skills. The participants were clarified, at various points in their development, about the diagnosis and its implications. Access to psychological counseling was also perceived as important at times when facing stressful events. All the mothers of the participants highlighted the importance of psychological support in the diagnosis phase and in the clinical treatment, broadening their understanding of the situation and enabling a better quality of communication with the healthcare staff.

None of the participants had a significant low score in WHOQOL-bref scale, which indicates they have a good quality of life in most domains.

5 FINAL REMARKS

The present study is composed of a few cases, preventing generalizing the results to other individuals with DSD. However, the method employed helps to understand the phenomenon of adolescence in individuals with DSD and female sex of rearing. The tests' results, as well as the data obtained through charts' review, enable a better understanding of this condition and its possible impacts in this age group. Transition to adult life is usually expected to be a challenge. For these participants this challenge seems to be more intense and difficult. With a childlike psychological configuration, they seem to avoid sexual

encounters with others and even with themselves, adopting a defensive and immature attitude.

These patients and their families need ongoing emotional and care support, as well as medical information related to the clinical condition. Communication is a key element to cope with the condition, allowing a smooth adaptation in society.

6 REFERENCES

Berenbaum, S.A. & Meyer-Bahlburg, H.F. (2015). Gender development and sexuality in disorders of sex development. *Hormone and Metabolic Research, 47*(5), 361-366. doi: 10.1055/s-0035-1548792.

Brito, A., Schaab, B.L. & Remor, E. (2019). Avaliação psicológica de pacientes com doenças crônicas não transmissíveis. In: *Avaliação psicológica nos contextos de saúde e hospitalar*. C.S. Hutz, D.R. Bandeira, C.M. Trentini, E. Remor (orgs.). Porto Alegre: Artmed, 150-159.

Cohen-Kettenis, P.T. (2010). Psychosocial and psychosexual aspects of disorders of sex development. *Best Practice & Research Clinical Endocrinology & Metabolism, 24*, 325-334.

Cools, M., Nordenström, A., Robeva, R., Hall, J., Westerveld, P., Flück, C. et al. (2018) Caring for individuals with a difference of sex development (DSD): a consensus statement. *Nature Review Endocrinology, 14*: 415-429.

Crouch, N. & Creighton, S.M. (2014). Transition of care for adolescents with disorders of sex development. *Nature Rewiews/Endocrinology, 10*, 436-442.

Freda, M.F., Dicé, F., Auricchio, M., Salerno, M & Valerio, P. (2015). Suspended sorrow: the crisis in understanding the diagnosis for the mothers of children with a Disorder of Sex Development. *International Journal of Sexual Health,* 27, 186-198.

Gardner, M. & Sandberg, D.E. (2018). Navigating surgical decision making in Disorders of Sex Development (DSD). *Frontiers in Pediatric, 6*, online article.

Gardner, M. & Sandberg, D.E. (2018). Navigating surgical decision making in Disorders of Sex Development (DSD). *Frontiers in Pediatrics* [online], *6*, (DOI: 10.3389/fped.2018.00339).

Hamed, S.A., Metwalley, K.A. & Farghaly, H.S. (2018). Cognitive function in children with classic congenital adrenal hyperplasia. *European Journal of Pediatrics, 177*, 1633-1640.

HEMESATH, T. Anomalias da diferenciação sexual: representações parentais sobre a constituição da identidade de gênero. Psicologia: Reflexão e Crítica, 2013. Disponível em: http://www.scielo.br/pdf/prc/v26n3/v26n3a18.pdf. Acesso em: 01 out. 2018.

Hemesath, T., de Paula, L.C.P., Carvalho, C.G., Leite, J.C.L., Guaragna-Filho, G. & Costa, E.C. (2019). Controversies on timing of sex assignment and surgery in individuals with Disorders of Sex Development: a perspective. *Frontiers in Pediatrics* [online]*, 6*, (DOI: 103389/fped.2018.00419).

Kleinemeier, E., Jürgensen, M., Lux, A., Widenka, P.M. & Thyen, U. (2010) Psychological adjustment and sexual development of adolescents with disorders of sex development. *Journal of Adolescent Health. 47*(5), 463-471. doi: 10.1016/j.jadohealth.2010.03.007.

Lee, P.A., Nordenström, A., Houk, C.P., Ahmed, S.F., Auchus, R., Baratz, A., Baratz, Dalke, K., Liao, L.M., Lin-Su, K., Looijenga, L.H., Mazur, T., Meyer-Bahlburg, H.F., Mouriquand, P., Quigley, C.A., Sandberg, D.E., Vilain, E., Witchel, S. and Global DSD Update Consortium. (2016). Global Disorders of Sex Development Update since 2006: Perceptions, Approach and Care. *Hormone Research Paediatrics, 85*(3), 158-180. doi: 10.1159/000442975.

LEVY, R. O adolescente. In: Eizirik,C.L.; BASSOLS, A.M.S. O ciclo da vida humana: uma perspectiva psicodinâmica. Porto Alegre: Artmed, 2013, p. 167-179.

Patton, G.C. & Viner, R. (2007). Pubertal transitions in health. *Lancet, 9*, 369-1130.

Schützmann, K., Brinkmann, L., Schacht, M., Richter-Appelt, H. (2009) Psychological distress, self-harming behavior, and suicidal tendencies in adults with disorders of sex development. *Archives of Sex Behavior, 38*(1), 16-33. doi: 10.1007/s10508-007-9241-9.

Schweizer, K., Brunner, F., Gedrose, B., Handford, C. & Richter-Appelt, H. (2017). Coping With Diverse Sex Development: Treatment Experiences and Psychosocial Support During Childhood and Adolescence and Adult Well-Being. *Journal of Pediatric Psychology*. Disponível em: https://www.ncbi.nlm.nih.gov/pubmed/27452605. Acesso em: 01 out. 2018.

Thyen, U., Ittermann, T., Flessa, S., Muehlan, H., Birnbaum, W., Rapp, M., Marshall, L., Szarras-Capnik, M., Bouvattier, C., Kreukels, B.P.C., Nordenstrom, A., Roehle, R., Koehler, B. (dsd-LIFE group). (2018). Quality of health care in adolescents and adults with disorders/differences of sex development (DSD) in six European countries (dsd-LIFE). *BMC Health Service Research, 18*(1): 527. doi: 10.1186/s12913-018-3342-0.

YIN, R. K. Estudo de caso: planejamento e métodos. Porto Alegre: Bookman, 2005.