



Case Study

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Floating Harbor Syndrome and / or Pellitier-Leisti Syndrome

René Schubert *

Corresponding Author: René Schubert, Clinical Psychologist (CRP 06/65624) and Psychoanalyst, Rua Eça de Queiroz, 386 – São Paulo/SP – Brazil.

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Abstract

Fewer than 50 cases of Floating-Harbor Syndrome are documented in the medical literature. Its cause is unknown. Most cases are sporadic, but some are hereditary, with the autosomal dominant transmission. The typical phenotype of the syndrome is the triangular face with a prominent nose. Short stature and delay in expressive language acquisition are commonly described. Also, some anomalies regarding tooth development and possible pictures of celiac disease. Puberty may come forward. There are some occurrences of (mild) intellectual disability, but the same is not noticed in all cases. This presentation aims to present a case report of the follow-up in psychotherapy of a young man who has the aforementioned syndrome. The psychotherapeutic treatment started when he was 6 years old in 2012 until the moment in 2017, and the description will be of this psychological follow-up with the patient, family, school and with other colleagues from other areas who are following the case. Psychometric assessments of intelligence have been carried out throughout these years and will also be presented.

Keywords: *Floating Harbor Syndrome, Pellitier-Leisti Syndrome, Case study, Psychodiagnosis, Play therapy, Psychotherapy.*

Introduction

Fewer than 50 cases of Floating-Harbor Syndrome are documented in the medical literature. Its cause is currently unknown. It is pointed out that cases are sporadic, but some are hereditary, with the autosomal dominant transmission. The typical phenotype of the syndrome is the triangular face with a prominent nose. Short stature and delay in expressive language acquisition described. Some anomalies regarding tooth development and possible celiac disease. The phenomenon of advancing puberty can occur. There are descriptions of cases in which intellectual disability is present, but the same is not noticed in all cases.

This case report aims to describe the psychotherapy follow-up of a young man who was diagnosed with Floating-Harbor Syndrome. The psychotherapeutic treatment began when he was 6 years old in 2012 and in this report, it will continue until 2017, and the description will be of this psychological follow-up with the patient, family, school and with other colleagues from other areas who are following the case. Psychometric assessments of intelligence have been carried out throughout these years and will also be presented.

The Leisti-Hollister-Rimoin or Floating-Harbor syndrome

Leisti-Hollister-Rimoin or Floating-Harbor Syndrome is classified in the ICD-10 as Q87.8. Your description in this: other syndromes with specified congenital malformations, not elsewhere classified.

This disorder is named after the hospital where it was first described, Boston Harbor Floating, by Pelletier and Feingold, in 1973 and then, in 1975, by J. Leisti and Collaborators, described the same disorder in a patient observed at General Hospital, California. The causes of this syndrome are unknown. There is a hypothesis that it may result from a new mutation (MELDAU, 2017).

It is a growth deficiency syndrome characterized by short stature, characteristic face and delay in speech development. Inheritance is possibly autosomal dominant. (LACOMBE, 1995).

Researchers WHITE, MORGAN and Collaborators (2010) in a study with 10 patients point out that the diagnosis of Floating Harbor Syndrome requires the typical facial dysmorphism associated with age, accompanied by abnormalities in the hands, speech disorder and behavioral phenotype. They highlight severe speech and language disorders that affect the exchange of information and written communication. In addition, there is a high probability of the presence of Attention Deficit Hyperactivity Disorder (ADHD) during school age. If precocious puberty occurs, puberty suppression should be considered to maximize height into adulthood. Bone age is consistently delayed when measured in early childhood but variable when measured in late childhood.

From the study of 52 individuals diagnosed with Floating Harbor Syndrome and carrying out the phenotypic study of the cases NIKKEL, DAUBER and Collaborators (2013) point to the characteristic facial gestalt. It is not uncommon for an individual with this syndrome to have additional anomalies and health complications that require medical intervention, comprehensive study and investigation in clinical evaluation, and further surveillance. In general, individuals with this syndrome are healthy and despite some deficiencies, enjoy a good quality of life.

In other studies, the incidence of a certain intellectual deficit was also reported: in all cases, each individual showed a variable degree of intellectual disability and learning difficulty, ranging from borderline normal to moderate intellectual disability. Also noticed in some cases are changes in dentition (small teeth and recurrent dental problems), visual impairment, celiac disease, genitourinary and cardiac disorders. In a few cases, there is an incidence of autism spectrum disorder and obsessive-compulsive disorder (MELDAU, 2017; LEISTI, HOLLISTER, RIMOIN, 1975).

The clinical history

In September 2012 the psychotherapist was approached by Mr. Yoko to carry out the cognitive assessment of his only child Yan, 6 years old at the time. The indication came from a colleague who knew the work of psychotherapists in a children's clinic and the experience with child psychodiagnosis and monitoring of special children (MENDES & SCHUBERT, 2006). The initial complaint was the intense agitation of the boy and a characteristic of swearing a lot.

Both parents, Yoko and Sam, came to the anamnesis consultation and reported their concerns about the evolution of their son Yan. They pointed out that it was discovered, after several investigations, the presence of a rare genetic Floating Harbor syndrome. The evaluations started right after the postpartum period, due to the difficulties in growing and gaining adequate weight. It was at the Hospital das Clinicas that a team of endocrinologists characterized the condition as a phenotype with growth deficit. Due to the lack of information about the syndrome and the situation presented by Yan, the parents sought help and guidance from health professionals.

At the time, the parents point out that this was a sometimes irritable child with a persistent behavior of swearing and screaming. Independent and autonomous, he made friends with ease and was known for his charisma. She already had an intense curiosity and sometimes manipulated her body and this worried her mother.

As for neuropsychomotor development, there were some delays – the first telltale factor was short stature and difficulty in gaining weight. The first words came at a year and a half. He was only two years old. She took off her diaper when she was three years old. She sucked on a pacifier and suckled on a bottle until she was three to four years old.

At school, he was in his first year and was described as a very agitated, restless and easily distracted child. Pointed out a gap in the pedagogical and educational issues. She had difficulty getting organized, doing her homework, and at home, she avoided talking about what happened at school.

At the first contact with the psychotherapist, he was receptive, participative and pleasant. Some cognitive and personality assessments were done with Yan. The materials and methods used were: H.T.P. (Van Kolck, Brazil, 1984), Design of the Family, Personality Traits Scale for children (Editora Vetor, Brazil, 2004), Raven's Colored Matrices (Editor Center for tests and research in psychology, Brazil, 1999), R2 - non-verbal and intelligence test for children (Editora Vetor, Brazil, 2000), Child Apperception Test with animal figures (CAT-A, Mestre Jou, Brazil, 1981) and Playful observation.

In his contact with toys and games, Yan demonstrated that his playing was healthy and age-appropriate, with a lot of fantasy and with the theme of good versus evil characters present. He always included the psychotherapist in the game and after the session showed the parents some of the things he had drawn, seen or done. When playing, Yan gave vent to his agitation in a more punctual way, despite being so anxious at times that he got in the way of what he intended to do. The playful is the child's language showing how the child is elaborating concepts of external reality, of the adult world (WINNICOTT, 1975). Through playing, the quality of Yan's bond and psychic flexibility can also be verified, as well as the presence of an organized psychic structure such as it would be in the case of the neurotic structure, discarding the presence of psychic instability, mood lability or disorganization as aspects of a psychotic structure – this factor needed to be verified since it was a syndromic condition that in the scientific literature describes some cases of comorbidities and associated psychiatric conditions. Just as looking at the psychic structure allows the psychotherapist to better elaborate the therapeutic posture, clinical strategies, referrals and prognosis, evolution of the condition (FREUD, 1909 and 1924).

Based on the psychodiagnostic evaluation, it was concluded that there was a clear presence of anxiety and psychomotor agitation. Positive intellectual development and sociability. The attachment and affective relationship were age-appropriate and Yan's curiosity encourages him to develop in several aspects. The presence of the syndromic picture and the symptoms presented pointed to the maintenance of psychotherapeutic follow-up with family guidance. Speech difficulties were already being monitored by a speech therapy team and a neurologist was monitoring the case.

Since the beginning of the treatment, the parents were very present in Yan's follow-up. The mother closely followed her pedagogical development and brought it to the psychotherapist his doubts about his daily dealings with Yan and his concerns about his sociability, discipline, expression of affection and relationships with family, friends and colleagues. The mother also updated the psychotherapist on the opinions, suggestions, examinations and conclusions of the other professionals who accompanied Yan, such as the neurologist, the speech therapist and the various teachers he had in subsequent years.

Follow-up with a speech therapy team occurred from Yan's 4 years old. The parents' perception is of significant evolution from oral language, written language and text interpretation.

Yan has been following up on Neurology since he was little. It was from 2014 onwards that Dr. M. Neurologist continued the follow-up. As for the syndrome presented, what drew a lot of attention was the psychomotor agitation and the damage it brought to the learning process. As a result of this, it was from 2012 that the psychopharmacological use of Ritalin began. From 2015 to 2017 he was medicated with Concerta (18mg). In 2017, the association of Sertraline (25mg) and Melatonin (3mg) was started. During these years, breaks were always made on the weekends and school holidays. This always had the supervision of the neurologist and follow-up in the office with the psychotherapist. The medication was aimed at better performance and school performance, and in this sense, it obtained good results with positive returns described by the teachers and school coordinators.

Cognitive assessments also corroborate that the use of medication helped in Yan's development, making him concentrate better and that he could develop strategies for learning.

Cognitive assessments: a progressive study from the clinical case

During these 6 years, intelligence and neuropsychological tests were applied to verify Yan's cognitive development and the effects of the medication. All tests applied are previously approved as intellectual and cognitive assessment tools by the Federal Council of Psychology (SATEPSI, 2017).

Applied cognitive and intelligence assessments: Nonverbal test of color matrices by RAVEN (Editor Center for Tests and Research in Psychology, Brazil, 1999); Non-Verbal Reasoning Test for Children - TNVRI (Editora Vetor, Brazil, 2005); R2-non-verbal test and intelligence for children (Ed. Vetor, Brazil, 2000); Nonverbal General Intelligence Test / TIG-NV (Casa do Psicólogo, Brazil, 2008)

Although several researchers point to the incidence of intellectual disability in Floating-Harbor Syndrome, this aspect was not verified in Yan. Since the psychodiagnostic assessment in 2012 through the Raven and R2 Yan tests, he presented intellectual performance within the mean for the chronological age presented. What always drew attention was her psychomotor agitation and as a result this recurrent inattention. However, with encouragement and follow-up, Yan was able to concentrate and perform the requested exercises, showing good results for concrete reasoning and evidence of the development of abstract reasoning and interpretation.

In psychotherapy, as these tests were applied and reapplied, Yan worked on strategies so that he could concentrate and keep focused on the exercises performed. One strategy he often repeats and reports helping is the ability to stretch, stretch, stretch, and engage in mindful breathing for a while. Sometimes he gets up, takes a turn and goes back to the exercise. It does this in a relaxed and light way and is satisfied with having a return on its positive results.

It was only in 2013 that the neuropsychological assessment using the TIG was inconclusive, as Yan demonstrated that he did not understand the complexity of the test. In other words, there was still no cognitive maturity for such testing. It was reapplied in 2016 when Yan understood what was expected in the test and showed results within the Brazilian average. Complex, abstract and interpretive reasoning is the greatest difficulty for Yan and as a result, he presents greater difficulties in subjects such as Portuguese and mathematics. This meant that joint work with the mother, teacher, speech therapy, neurology and psychotherapy were intensified to help Yan in the best possible way in this regard.

Tests were repeated in 2015, 2016 and 2017, showing the maintenance of intellectual results within the average for the Brazilian population. In this sense, variations in intelligence levels were not noticed. It was noticed that Yan was maturing his reasoning and skills, perhaps at a different time from his classmates, but even so, in continuous development. In this sense, the encouragement given by parents and professionals who accompanied him was fundamental.

Therapeutic bond and affective development

Yan has a fascination for heroes. From the first moment in the office, he brought to the psychotherapist stories of his favorite heroes. At the time it was Ben 10, from this we went to Batman, X-Men, Superman, Spider-Man, Hulk, Iron Man, Yu Gi Oh, Naruto and so many others. This is the point – so many others – Yan has always shown flexibility and movement in his search for models. This provided important input for the psychotherapist and this interest and search for heroes was stimulated and worked on in the individual psychotherapeutic environment of the office. Nothing better than good models, especially if they allow molding, flexibility, change and adaptations.

Through these heroes, we enter different worlds such as graphics, folding, crafts, memory and board games, music, reading and even, of course, lessons and school assignments. These heroes were bridges from playful to adult – from play to responsibility, of course, always using language and resources close to Yan. Heroes were good models of discipline and dedication and showed both positive and negative aspects, namely, otherness. Heroes had different clothes and habits. Appropriate and other strange behaviors for now. For example, Hulk demonstrated the consequences of outbursts of rage and lack of control, while Tony Stark, being dedicated and intelligent, built for himself an iron armor that could fly. And Yan identified with these heroes for a while. Designed, built and produced clothing and accessories. He sang song lyrics and told the psychotherapist about particulars he had discovered. And because the psychotherapist is interested in these matters and enters this universe together with Yan, the bond became healthy and one of continuous exchange.

In this way, Yan allowed the introduction of new stimuli, games, practices without resisting – he trusts that it will develop and that it will be fun. And the psychotherapist always seeks to focus on their potential and abilities. The focus is exactly on the child's potential, regardless of the diagnosis – such a cut allows for the healthy and spontaneous appearance of social and cognitive skills (SCHUBERT, 2008).

And Yan maintains an open and affective posture in contact and social exchange. It respects the limits imposed by adults and it is noticed that it quickly responds to commands, requests made either by the mother or father, for which it shows great affection and respect.

For parents, the family guidance space proved to be very important to provide security and help them with doubts and anxieties related to the child's development. Many dynamics and behaviors presented by Yan, when reflected and translated by the psychotherapist, brought relief and direction to the parents. As postulated by WINNICOTT (2001): "In the psychological universe, there is a tendency to development that is innate and that corresponds to the growth of the body and the gradual development of certain functions", there is a lot of anxiety about the development of these functions and healthy and adequate growth of Yan, and in this meaning, psychotherapeutic accompaniment provides a cutout, an open space for construction and dialogue. This benefits Yan, his family and the continuity of psychotherapeutic accompaniment as a creative and safe space for the manifestation of psychic dynamics and potential.

Conclusion

One could say what is concluded so far on this journey. Yan's characteristic of compulsively swearing diminished until it ceased. Motor agitation is a more conscious factor for Yan and strategies were developed to overcome the difficulties and obstacles generated by this characteristic. Your body awareness is increasingly present, bringing the notion of movement and its consequences.

Difficulties at school are a stimulus to seek alternatives. We don't focus on the problem in the office, but on the solution – on the skills and talents available in Yan to achieve the expected results. Yan's emotional and social intelligence stand out in this respect, often covering up and minimizing her difficulties and resistances. However, helping him to become aware of these difficulties and working with patience and persistence, Yan is increasingly moving towards overcoming these difficulties. The look and monitoring of various knowledge and professionals were made available to Yan by his parents so that he could move forward. And so it goes on, with confidence.

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