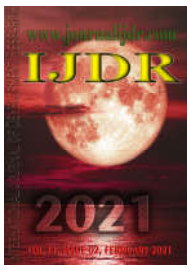
ISSN: 2230-9926

Available online at http://www.journalijdr.com

IJDR

International Journal of Development Research
Vol. 11, Issue, 02, pp.44346-44349, February, 2021
https://doi.org/10.37118/ijdr.21183.02.2021



RESEARCH ARTICLE OPEN ACCESS

ROBINOW SYNDROME: MEDICAL AND STOMATOLOGICAL CONSIDERATIONS FOR COMPREHENSIVE PATIENT MANAGEMENT

Cristhiane Olivia Ferreira do Amaral*[1], Davi de Alencar Gonçalves[2], Luís Augusto Rossi Nery[2], Vinicius Carvalho Rodrigues[2], Mariana Olívia Ferreira do Amaral[3], Eliane Cristina Gava Pizi[4] and Fabiana Gouveia Straioto[4]

[1]DDS, Ms, PhD. Department of Special Care Dentistry and Pediatric Dentistry, Dental School, University of West São Paulo, Presidente Prudente, Sao Paulo, Brazil; [2]Dentistry, Dental School, University of West São Paulo, Presidente Prudente, Sao Paulo, Brazil; [3]Graduate student, Medical School, University of West São Paulo, Presidente Prudente, Sao Paulo, Brazil; [4]DDS, Ms, PhD, Professor Dental School, University of Western São Paulo, Presidente Prudente, Sao Paulo, Brazil

ARTICLE INFO

Article History:
Received 21st December, 2020
Received in revised form
14th December, 2020
Accepted 08th January, 2021
Published online 24th February, 2021

Key Words:
Syndrome, Multiple Abnormalities, Oral Health, Dental Assistance.

Corresponding author:
Cristhiane Olivia Ferreira do Amaral

ABSTRACT

Introduction: Robinow's syndrome (RS) is a rare, skeletal malformation occurs that can also affect other organs, t in a wide spectrum of clinical characteristics, which can be: craniofacial, dental, musculoskeletal, urogenital and presence of systemic complications. **Objective:** to make considerations about systemic alterations, to identify the general characteristics, oral diseases in Robinow Syndrome and additionally to report a clinical case of a patient with this syndrome, highlighting aspects of dental interest. **Case Report:** It was observed in the evaluation and monitoring of the patient the following characteristics general: short stature, fetal face, exophthalmos, hypertelorism, cognitive impairment, skeletal abnormalities, renal impairment, cognitive impairments. The patient is followed up dentally since he was a baby and the following oral characteristics were present: malocclusion, deep palate, crowding, unilateral posterior crossbite, gingival hyperplasia, difficulty in cleaning, nausea during the dental approach, but with satisfactory quality of oral health op atient reported is male, 12 years old, with diagnosis of S R. **Conclusion:** Approach and comprehensive treatment should be encouraged, and monitoring by a multiprofessional team, as these individuals have complex problems in relation to systemic, behavioral and dental characteristics. The Early dental care to these patients should be performed so that do not require complex and invasive dental treatments due to systemic and cognitive impairment.

Citation: ***Cristhiane Olivia Ferreira do Amaral, Davi de Alencar Gonçalves, Luís Augusto Rossi Nery, Vinicius Carvalho Rodrigues, Mariana Olívia Ferreira do Amaral, Eliane Cristina Gava Pizi and Fabiana Gouveia Straioto,*** **2021.** "Robinow syndrome: medical and stomatological considerations for comprehensive patient management", *International Journal of Development Research*, 11, (02), 44346-44349.

INTRODUCTION

Robinow syndrome (RS) was described in 1969 in the American Journal of Diseases of Children by the authors Meinhard Robinow (geneticist), Frederic N. Silverma n and Hugo D. Smith both doctors. This syndrome is rare genetic disorder[1] and the prevalence is unknown. The incidence reported is 1: 500,000 births[2]. The frequency of incidence in male and female is equivalent, but it is reported to be higher in males. The low prevalence may be justified, due to percentage of 5-10% of the infant death with cardiac problems due to syndrome[3]. The diagnosis of this syndrome is determine using genetic testing of the ROR2 gene matching, and association with clinical characteristics[4] Prenatal diagnosis is possible from the 19th week of pregnancy by fetal ultrasound analyzing the long bone length[5].

The Robinow syndrome is a syndrome that express morphological characteristics: abnormal morphogenesis of the face (craniofacial, buccal), genitals (urogenital) and vertebral segmentation (musculoskeletal)5 and systemic complications[6]. The cranial facial manifestations frequently are fetal face, macrocephaly, eyelid clefts, hypertelorism, lymph extinction, ethmoidal hypoplasia and frontal sinus, sphenoid and maxillary, flattened nasal bridge with open nostrils, prominent forehead, low implanted ears and with pavilion auricular presenting deformity[6]. The patient with Robinow syndrome also has some oral specific characteristics: upper lip appearance of inverted link as "V" exhibiting the incisors and gingival tissue, long philtrum of upper lip, gingival hyperplasia, crowded teeth, malocclusion, tooth eruption delayed, micrognathia, macroglossia, ankyloglossia, benign migratory glossitis, deep palate, bifid uvula, cleft lip and palate in 9% of individuals with RS[7].

The musculoskeletal manifestations presented are moderate short stature in the majority of cases. Mesomelic branchimelia, which is characterized by the shortening of the middle portion of the upper limbs due to anomaly in the growth of the radius and ulna. It is found in most of those affected and in some cases it is demonstrated only through radiological measurements. The phalanges of the fingers are short and the distal phalanges of the thumbs can be bifid. In 85% of cases are observed small hands with syndactyly, clinodactyly and 5th finger brachydactyly. There are more serious vertebral abnormalities that this patient could presents such as fusion of vertebrae, progressive scoliosis and ribs anomalies[2,6,7.] The urogenital manifestations already present at birth. The genitals are abnormal, especially in males, small penis and scrotum and can be also have cryptorchidism8. Patients with RS syndrome may suffer congenital cardiac problems, and although there is no clear pattern. The most common conditions include pulmonary stenosis and atresia. Congenital heart defects are the princi parents because death untimely 9. Intelligence may be normal, but patients may experience developmental delays, as well as seizures[10].Others abnormalities that can affect theses patients are reported in the literature kidney, hydronephrosis that might lead to urinary tract infections11. Additionally, these individuals may suffer from cystic kidney dysplasia[12]. The treatments of RS patients are symptomatic and corrective surgery like as syndactyly repair, severe scoliosis, cleft lip and cleft palate[13–16]. The Robinow syndrome is rare and poorly studied especially considering the dental characteristics. The dental treatment of this patients request specific knowledge about possible oral and systemic alterations and some especial care. The aim of this case report is discuss about general, systemic and oral characteristics of patient diagnosed with Robinow syndrome.

CASE REPORT

This case report descriptive of a GFL patient, male, Brazil, 12 years old, born and living in Presidente Prudente – SP. The patient has molecular diagnostic of Robinow syndrome. The responsible for the patient presented in this case report was informed of the purpose and about the methods that will be used in this study. The responsible for the patient signed the Informed Consent and Informed that was based upon the specific ethical aspects according to the originating principles of Helsinki Declaration. The patient was referred to the Dental Specialties Center to perform the dental treatment, due to be bearer of Syndrome Robinow. This patient had been monitoring and under dental prevention procedure since 2 years old. The dental prevention protocol is cleaning procedure associated with fluoride topical application every 4 months since then. During the anamnesis interview, the mother reported that the diagnosis of the syndrome was established at 6 months of age. The diagnosis of the syndrome was obtained by genetic testing sequencing of the ROR2 gene, associated with clinical caracteristics that the baby presented in the perinatal period. He is only child, his prenatal medical history reported when ate the sixth month of pregnancy the baby was diagnosed with polycystic kidney. He was born by normal childbirth, weighing 3.954 kg and 51 cm length. The postnatal medical history reported by mother. She described that the baby was submitted to bronchial aspiration of amniotic fluid and he had to be hospitalized during10 days under medical care. The mother related that the baby did not cry, and he had generalized muscle hypotonic. After 6 months of the birth, based on physical and systemic characteristics that led to a clinical suspicion of the syndrome the laboratory exam was requested for the diagnosis of Robinow Syndrome. The general characteristics are: 1.45m high and 58 kg. His body mass index - BMI is 27.6, he is considered overweight. The blood pressure checked was 130/90 mm Hg and the heart rate was 70 beats per minute. The eyes of the patient are very prominent (Figure 1) as kwon exophthalmos, but this eye projection in Robinow syndrome differs from true exophthalmos. Therefore, the eyes do not protrude from the orbit, the in Eve I. Addionatlly, there is a deficiency the lower eyelid which gives the eyes more prominent appearance17. This pseudo exophthalmia may require surgical correction, which has not been performed yet, because the eyes can not close completely leading to ocular constant hyperemia.

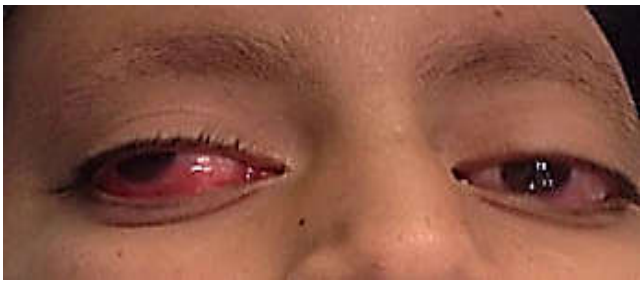


Figure 1. Image of the eyes of the patient with prominence associated with deficiency the lower eyelid to close completely leading an ocular constant hyperemia

The patient has renal impairment from birth and present stenosis junction ureteral pielo (JUP), is a congenital narrowing the place where is the junction between the renal pelvis and the ureter[18].

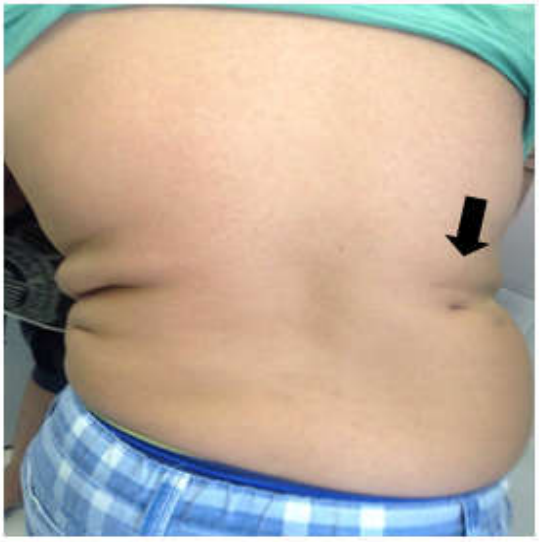


Figure 2. The arrow indicate the scar from surgeries performed for nephrectomy and pyeloplasty

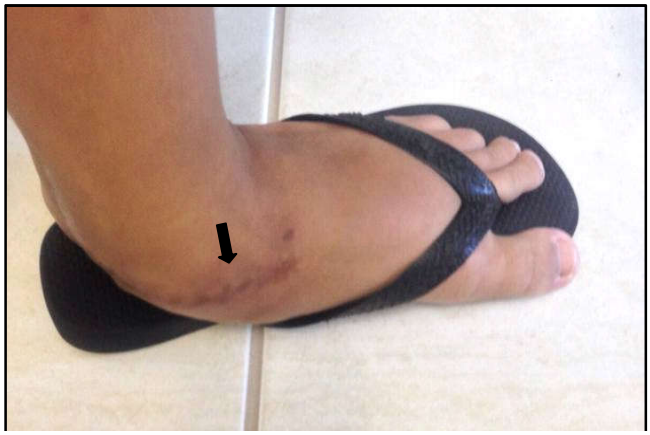


Figure 3. The arrow indicate the scar of the foot patient with scar from the surgeries performed to correct his gait, the flat feet, ligament laxity

The cognitive and mental developing presented deficit. The neurological development has been slow, but positive, with the acquisition of new milestones engines. He had a delay in his speech process and walk, both he started just after 4 years of age. Others deficits were reported by his mother: ride bike skills, fine motor skills, emotional immaturity, socialization difficult (he do not have capacity to go out alone), hyperactivity that it was perceived during dental treatments. The patient has been taking the risperidone medication since 4 years old, that it is an antipsychotic used for behavioral control[19].

The general characteristic were: stunting light without shortening of the limbs, scoliosis, joint subluxation radioulnar because of ligament laxity, cryptorchidism, congenital heart murmur, abnormality in kidney tract, hydronephrosis, and cystic dysplasia kidney. The cranial facial and stomatological manifestations observed were fetal face, prominent forehead, eyelid clefts, hypertelorism, exophthalmos, low ears implantation (Figures 4a and 4b), upper lip with an inverted "v" appearance exposing the incisors and gums of the upper lip, atypical swallowing and oral breathing.

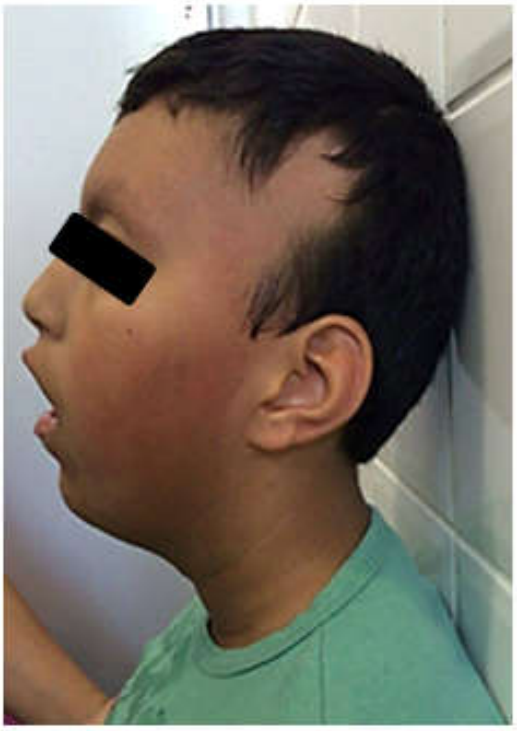


Figure 4a: Profile view of patient

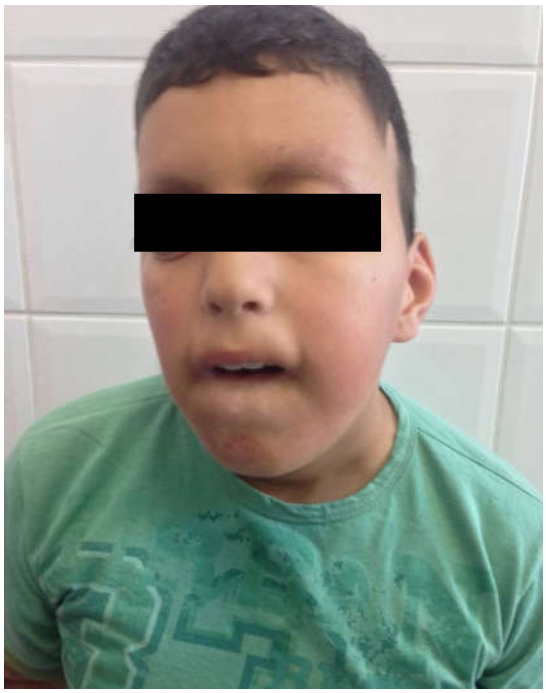


Figure 4 b: Front view

The intraoral examination observed the presence of mild gingival hyperplasia associated with gingivitis (Figure 5), deep palate and any caries cavity was found (Figure 6). Considering the mother is responsible for all the daily care of the child life the education about dental was realized to the caregiver. Instruction of how to brush his teeth, reduced of sugar intake.

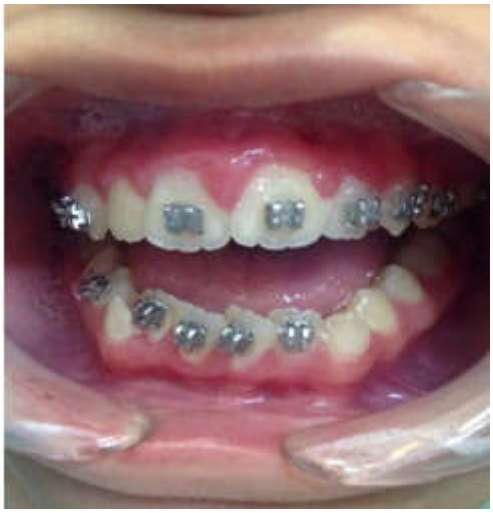


Figure 5. Front view, there are presence of mild gingival hyperplasia associated with gingivitis

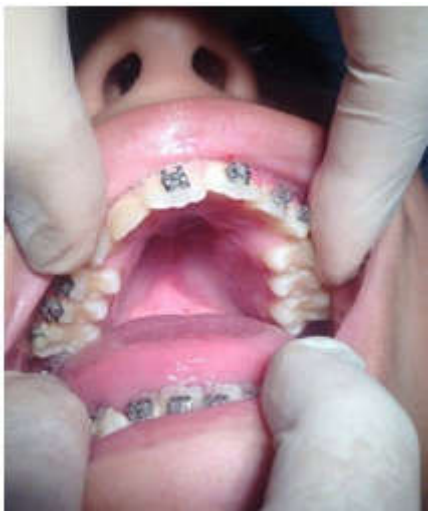


Figure 6. Intra buccal view, deep palate and any caries cavity was found

During dental treatment the patient was partially collaborate. He accepted to take a panoramic radiograph with some difficulty. He did not like the light of reflector. However, he allowed the use of prophylactic paste, robson brush and micromotor. His mother was present in all of the treatment sessions (Figure 8).

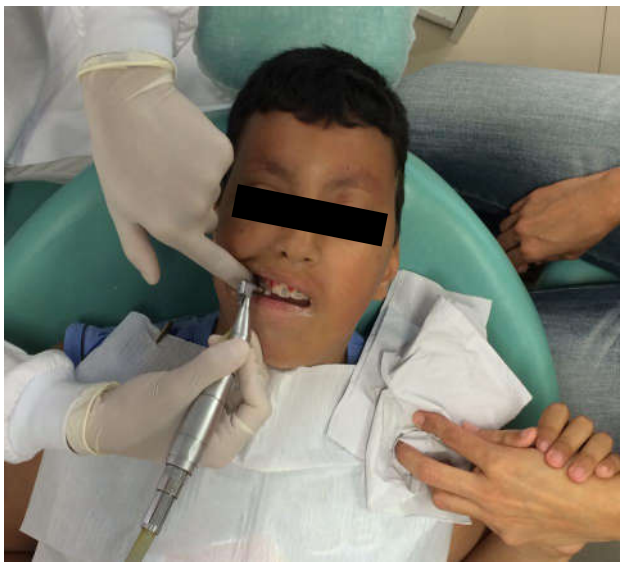


Figure 8: Image of patient during dental treatment procedure. The mother of patient stayed together holding the hand of patient during all dental treatments appointments

The analyses of radiograph exam, panoramic radiography image (Figure 9) showed presence of supernumerary teeth. It was observed short space for 17 and 27 teeth, causing retention of those teeth.

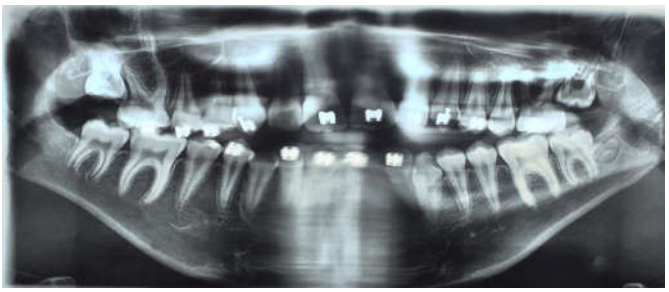


Figure 9. Panoramic radiograph image. Retention of teeth 17 and 27

DISCUSSION

The hypothesis of study was that patients who are diagnoses carrying genetic syndromes have unmet needs for dental approach related to hygiene dental. The behavioral barrier, and systemic characteristics can interfere in the treatment plan. Continuous use of controlled drugs and salivary aspects lead them to poor quality oral health, hence the need to seek dental prevention since baby. Robinow's syndrome is a genetic disorder with the highest number of diagnosed boys. In this study, we report a case of a 12-year-old boy who has attended a dental prevention program since he was a baby. In addition to requiring the establishment of frequency habits to the dental surgeon create routine and bond, generate care with oral hygiene, reducing the presence of caries and other oral diseases [20]. This syndrome is rapid medical suspicion, due to the variability of clinical and systemic characteristics that the patient may have [17], mainly renal impairment and generalized muscle hypotonia. The syndrome Robinow has systemic characteristics important to be observed by dentist. The case reported described the patient has renal impairment which is presented in 25% of individuals with RS, and also had a heart murmur which affects 13% of patients with the syndrome [2, 6]. These systemic condition must have been observed, as they influence the dental approach, as they require pre, peri and post invasive treatments, mainly surgical[21]. Cardiac abnormalities can be an important cause of morbidity and mortality in these patients [16]. The musculoskeletal manifestations presented by the patient, we only find scoliosis that is presented in 50% of patients with RS [2,6,7]. Bone age is delayed by 35 to 44% of RS cases, but the patient in question is not bone age delayed by radiographic wrist examination. The other musculoskeletal changes were not observed, only surgeries were performed on the patient's feet to correct gait [2,6,7].Regarding the cranial facial manifestations, the patient presented a fetal face which is the most pronounced feature of RS. Additionally, also presented eyelid clefts which is found in 88% of cases, which is found to be hypertelorism in 100% of the cases, exhilaration that is reported in 88% of patients, a prominent forehead that appears in 96% of patients and ears with low implantation, also reported in the literature [2, 6]. People with this syndrome also have a variety of oral characteristics: upper lip with an inverted “v” appearance exposing the incisors and the upper rim gingiva, triangle-shaped mouth. This characteristic is present in 94% of individuals with RS, also introduced into the patient in question will. The patient in this case presented the teeth crowding, poor tooth position, as well as 96% of the RS of patients in general, the palate of the patient was observed relatively deep in the same way that 80% of cases of RS. The presence of gingival hyperplasia was also investigated, which is also reported in 59% of patients affected by the syndrome, although this hyperplasia seems to be related to the use of the fixed orthodontic appliance [2,4,6,7,16,17]. Daily oral hygiene is reported by the mother as complicated, so the guardians must be properly instructed for its performance. They must be informed about the importance of carrying it out. If performing at home is very difficult or impossible, periodic return visits to the dental office should be scheduled as close as possible. The frequency of returns should vary according to the patient's caries risk / activity and the difficulty of cleaning at home [21].

CONCLUSION

Patients with Robinow Syndrome present diversity of general and oral characteristics. They have difficulty in oral hygiene due to the possibility of delayed neuropsychomotor development. Thus, it is important to provide early dental care for these patients, so that they do not need complex and invasive dental treatments due to systemic and cognitive impairment. The approach and comprehensive treatment must be carried out by a multidisciplinary team, as these individuals have complex problems in relation to systemic, behavioral and dental characteristics.

REFERENCES

1. Brunetti-Pierri N, Del Gaudio D, Peters H, Justino H, Ott CE, Mundlos S, Bacino CA. Robinow syndrome: phenotypic variability in a family with a novel intragenic ROR2 mutation. Am J Med Genet A. 2008; 146 (21): 2804-9.
2. Kim CA, Sugayama SMM, Ribeiro EM, Hajaj G C. Robinow syndrome. Pediatrics (São Paulo) 1994; 16 (4): 190-3.
3. Van Bokhoven H, Celli J, Kayserili H, van Beusekom E, Balci S, Brussel W, Skovby F, Kerr B, Percin EF, Akarsu N, Brunner HG. Mutation of the gene encoding the ROR2 tyrosine kinase causes autosomal recessive Robinow syndr ome. Nat Genet. 2000; 25 (4): 423-6.
4. Beiraghi S, Leon-Salazar V, Larson B, John M, Cunningham M, Petryk A, Lohr J. Craniofacial and intraoral phenotype of Robinow syndrome forms. Clin Genet. 2011; 80 (1): 15-24
5. Guven MA, Batukan C, Ceylaner S, Uzel M, Ozbek A, Demirpolat G. Prenatal and postnatal findings in a case with the autosomal recessive type of Robinow s yndrome. Fetal Diagn Ther. 2006; 21 (4): 386-9.
6. Hervert L TG, Loa Urbina ML. Atención Stomatological del pediatric patient con Robinow syndrome. ArchInvMatInf2013; 5 (2): 84-8.
7. Cerqueira DF, Souza IP. Orofacial manifestations of Robinow syndrome: a case report in a pediatric patient. Oral Surg Oral Med Oral Pathol Oral RadiolEndod. 2008; 105 (3): 353-7.
8. Wilcox DT, Quinn FMJ, Ng CS, Mireaux-Dicks C, Mouriquand PDE. Redefining the genital abnormality in the Robinow syndrome. J Urol. 1997; 157 (6): 2312-4.
9. Bacino C, Pagon RA, Bird TD, Dolan CR, Stephens K, Adam MP. Robinow ROR2-Related Syndrome. GeneReviews - Baylor College of Medicine; 2011.
10. Nareja MP, Gomez VL, Valde BJL. Robinow syndrome. Avg. Hosp. Infant. Mé.1988; 45 (8): 537-42.
11. Tufan F, Cefle K, Türkmen S, Türkmen A, Zorba U, Dursun M, Öztürk S, Palandüz S, T Ecder, Mundlos S, Corno D. Clinical and molecular characterization of two adults with autosomal recessive Robinow syndrome. Am J MedGenet A. 2005; 136 (2) : 185-9.
12. Soliman AT, Rajab A, Alsalmi I, Bedair SM. Síndromerecessivo RecessiveRobinow syndrome: with emphasis on endocrine functions. Metabolism. 1998; 47 (11): 1337-43.
13. Jones KL, Smith P. Patrones reconocibles of human malformations. 6th ed. Madrid, Spain: Elsevier Saunders; 2007.
14. Castells S, Chakurkar A, Qazi Q, Bastian W. Robinow syndrome with growth hormone deficiency: treatment with growth hormone. J Pediatr Endocrinol Metab. 1999; 12 (4) : 565-71
15. Sleesman JB, Tobias JD. Anesthetic implications of the child with Robinowsyndrome. Pediatrician Anaesth. 2003; 13 (7) : 629-32.
16. Soman C, Lingappa A. Robinow Syndrome: A Rare Case Report and Review of Literature. Int J ClinPediatrDent 2015; 8 (2): 149-152.
17. Patton MA, Afzal AR. Robinow syndrome. J Med Genet. 2002; 39: 305-10
18. Rim Molina CAF, Fancicani I, Muglia VF, Araújo WM, Cassini MF, Tussi JR S. Postnatal evaluation of intrauterine hydronephrosis due to ureteropelvic junction obstruction. Acta Cir Bras. 2013; 28 (1): 33-6.
19. Menezes GA, Fontenelle LF, Mululo S, M Versiani. Treatment-resistant anxiety disorders: social phobia, generalized anxiety disorder and panic disorder. Rev. Bras. Psychiatrist. 2007; 29 (2) 55-60.
20. Gudkina J, Brinkmane A.The impact of salivary mutans streptococci and sugar consumption on caries experience in 6-year olds and 12-year olds in Riga. Stomatologija. 2010; 12 (2): 56-9.
21. Amaral COF, Logar GA, Parisi AG, Takahashi K, Straioto FG. General and Stomatologic Aspects of Bardet-Biedl Syndrome .2014; 25 (6): 575-8.

****