Pickardt syndrome

Mayana Cerqueira Martins da Silva
Academic of medicine, by the Institution Universidade Salvador (UNIFACS)
Institution: UNIFACS
Address: Rua Minas Gerais, 298- apto 103. Pituba - Salvador, BA, CEP: 41830-020
E-mail: maycerqueirams@gmail.com

Alfredo Guerra Netto
Doctor, Morgana Potrich College (FAMP)
Goiânia City Hall - Emergency Management, Paço Municipal
Address: Rua T52, 180, Bairro Setor Bueno - Goiania, Goias, CEP: 74215-220
E-mail: netto_31@hotmail.com

Camila Cardoso Barbosa
Bachelor of Medicine from the University Center Athens - Uniatenas
Institution: Hospital São Rafael - Imperatriz Maranhão
Address: Avenida Bernado Sayao, n3600, Bairro três poderes, Imperatriz, Maranhão, CEP: 65903-250
E-mail: mimicamila@hotmail.com

Giovana de Heberson Souza
Academic of Medicine at the Evangelical University of Goiás- Campus Anápolis
Institution: Evangelical University of Goiás
Address: Al. das Rosas, 847, St. Oeste - Goiânia- GO, CEP: 74110-060
E-mail: giovanahs97@gmail.com

Gabriela Alves de Lima
Resident Physician of General Medical Practice by Universidade Estadual Paulista (UNESP)
Institution: Universidade Estadual Paulista "Júlio de Mesquita Filho"
Address: Rua Reverendo Francisco Lotufo, 663, apto 203. Vila Nogueira - Botucatu - SP, Zip Code: 18606810
E-mail: gabrielalima95@hotmail.com

Fernanda Durães Souto Rocha
Resident Physician in Family and Community Health Medicine at the University Hospital of Montes Claros (UNIMONTES)
Institution: State University of Montes Claros
Address: Avenida Cula Mangabeira, 562, Bairro Santo Expedito -Montes Claros-MG, CEP: 39401001
E-mail: nandasoutok@gmail.com

Isadora Leão Amuy
Doctor from the State University of Mato Grosso do Sul
Institution: State University of Mato Grosso do Sul
Address: Rua Dr.Werneck, 41, Bairro Vila Albuquerque - Campo Grande - MS,
ZIP Code: 79060-300
E-mail: isadoraleo@hotmail.com

Alessandra Andrade Lopes
Bachelor of Medicine from Centro Universitário de Brasília - UniCEUB
Institution: Hospital Municipal Alfredo Abrahão - Anápolis, GO
Address: Rua P-32, Quadra 21, Bairro Jardim Progresso - Anápolis, Goiás, CEP: 75063-610
E-mail: med.alessandralopes@gmail.com

Beatriz de Oliveira Onório
Graduated in Medicine from the Pontifical Catholic University of Campinas
Institution: Pontifical Catholic University of Campinas
Address: Santo Cristo, Bairro Recanto Verde - Carapicuíba, SP, CEP: 06351-280
E-mail: beatriz.onorio@gmail.com

Bruno Martin Leal
Bachelor of Medicine, by the Institution Faculdade Morgana Potrich - FAMP
Institution: UPA Araxá - MG
Address: Av. Pref. Aracely de Paula, Bairro Centro - Araxá, MG, CEP: 38184-022
E-mail: leal_351@hotmail.com

Eduardo Siqueira Borges
Graduating in Medicine at Faculdade Morgana Potrich - FAMP
Institution: Morgana Potrich College - FAMP
Address: Rua Fortaleza,355, Bairro Alto da Glória- Goiânia, GO, CEP: 74815710
E-mail: dudusikera@gmail.com

Gabriel Rodrigues Santos
Graduated in Medicine from the University of Rio Verde - UNIRV
Institution: Hospital Presbiteriano DR. Gordon - Rio Verde
Rua Alameda Amazonas, 1, Sector Morada do Sol-Rio Verde, GO, CEP:75909-035
E-mail: gabriels.7@hotmail.com

Heitor da Mata Xavier
Graduated in Medicine from ITPAC - Porto
Institution: ITPAC - Porto
Address: Avenida Castelo Branco, S/N, Bairro Centro - Silvianópolis, TO, CEP:77580-000
E-mail: heitorxavier96@hotmail.com

Isadora Souza Mendonça
Bachelor of Medicine by the Institution Faculdade Morgana Potrich - FAMP
Institution: UPA Trindade - GO
Address: Av. Trindade - St. Soares, Trindade - GO, CEP: 75380-000
E-mail: isadorasouzamendonca@gmail.com

Geovanna Rodrigues de Oliveira
Bachelor of Medicine, by the Institution Faculdade Morgana Potrich - FAMP
Institution: Cais Parque das Amendoeiras - GO
Address: Av. T15, n1222, Setor Nova Suíça, Goiânia - GO, CEP: 74230-010
E-mail: gevannarod4@gmail.com

Gabrielle Santiago Silva
Bachelor of Medicine, by the Instituto Master de Ensino Presidente Antônio Carlos - IMEPAC Centro Universitário Araguari
Institution: UPA Caldas Novas - GO
Address: Rua Gonçalves Dias, 76, Bairro Bosque - Araguari-MG, 38440-120
E-mail: gabisantiagos@hotmail.com

Gabriella Ribeiro Cabral de Oliveira
Bachelor of Medicine from Centro Universitário do Planalto Central Apparecido dos Santos - UNICEPLAC
Institution: Hospital Municipal do Céu Azul - Valparaíso - GO
Address: Q.55, Lot 01/04, Central Sector - Gama, DF, CEP: 72405-550
E-mail: gabicabral1521@gmail.com

Geovanna Rodrigues de Oliveira
Doctor by Faculdade Morgana Potrich – FAMP
Institution: Instituto Pedro Miranda - IPM
Address: Av T5 n1200 apto 304 Setor Bueno Ed Bougainville, Goiânia - Goiás Zip Code: 74230-042
E-mail: geovannarod4@gmail.com

Ingryd Inácio Ferreira Mesquita
Education: medical student
Institution: uniderp Anhanguera Campo Grande
Address: Rua da graciosa, número 61, bairro tiradentes, condomínio liv cidade jardim, torre 3 apto 104, Campo Grande MS, CEP: 79041-022
E-mail: medingrydinternato@gmail.com

João Nikolai Vargas Gonçalves
Graduating in Medicine from the University of Rio Verde – UNIRV
Institution: University of Rio Verde – UNIRV
Address: Sqs 113 bloco f apt 203, Brasília - DF CEP: 70376-060
E-mail: joaonikolai@gmail.com

Gabriela Coelho de Oliveira
Graduating in Medicine at Faculdade Morgana Potrich - FAMP
Address: Morgana Potrich College - FAMP
Address: Rua Armando Fajardo, 492, Bairro Vila Aurora- Rondonópolis, MT, CEP:78740-036
E-mail: gabrielacoelhodeoliveira@gmail.com

ABSTRACT

Introduction: The Pickardt-Fahlbusch syndrome, very rare, is found in the group of endocrine syndromes of interruption of the pituitary nail, resulting in tertiary hypothyroidism. The pathophysiology is not fully elucidated, suggesting alterations of chromosomal orders. Case presentation: A 13-year-old female patient, born in Gama - DF, was admitted to the emergency room of the Santa Maria Regional Hospital (HRSM) with decreased level of consciousness associated with severe hypoglycemia, showing improvement after reversal of hypoglycemia. Discussion: There is a higher prevalence of Pickardt-Fahlbusch syndrome in males, with symptoms of severe hypothyroidism well present, requiring complementary tests: laboratory and imaging. Once other causes are ruled out and diagnosis is made, therapeutic measures should be initiated early, reducing the effects and complications of the absence of thyroid hormones, especially in the

pediatric age group. Other characteristics may also be accompanied, masking the diagnosis, since alteration occurs at the level of the pituitary nail, which induces other symptoms. Conclusion: It is essential, given the various characteristics presented, to understand and study more about Pickardt's syndrome.

Keywords: Pickardt syndrome, Rare syndrome, Tertiary hypothyroidism.

1 INTRODUCTION

Pituitary nail interruption syndrome is a rare anomaly characterized by the presence of a thin or absent pituitary peduncle, associated hypoplastic or aplastic anterior pituitary, and ectopic posterior pituitary (PPE). It was first reported by Fujisawa *et al. in 1987 (FUJISAWA I et al, 1987*). It has a higher prevalence in males with a male-to-female ratio between 2.3 and 6.9:1 (TAUBER M et al, 2005).

The pathophysiology is not fully known, however, it is observed the association of the picture with the presence of chromosomal microdeletions, perinatal asphyxia in addition to genetic and environmental factors of pregnancy as possible causes. (WANG W et al, 2015).

Pickardt syndrome is characterized by tertiary hypothyroidism that occurs due to disruption of the portal veins between the hypothalamus and pituitary adeno. It is one of the variations of Pituitary Rod Interruption Syndrome and is marked by deficiency of thyroid-stimulating hormone, hyperprolactinemia and other deficiencies of pituitary hormones (GUTCH, 2014).

The clinical picture is marked by hormonal deficiencies generated by congenital hypopituitarism and in childhood it is possible to observe growth retardation, as well as short stature, seizures and hypotension. Delayed puberty may also occur. (R REYNAUD, 2011). It is possible to observe that the increased incidence of this disease is due to advances in radiology, since the use of magnetic resonance imaging in patients with suspected hypopituitarism increases the likelihood of early diagnosis and the initiation of treatment. (WANG W et al, 2015).

Through this study we seek to evaluate the etiology, pathophysiology, clinical picture, diagnosis and treatment of Pickardt Syndrome so that patients are effectively conducted, avoiding diagnostic errors and preventing complications arising from late treatment. Presentation of the case FRS, 13 years old, female, born in Gama - DF, was admitted to the emergency room of the Regional Hospital of Santa Maria (HRSM) with decreased level of consciousness associated with severe hypoglycemia, showing improvement after reversal of hypoglycemia. She reports recurrent hypotension and hypoglycemia. It has short stature for age and delayed pubertal development. It brings laboratory tests that showed a decrease in TSH and hyperprolactinemia. She was hospitalized for diagnostic elucidation, and magnetic resonance imaging of the skull was requested, which showed: hypoplasia of the adenohypophysis and ectopia of the neuropituitary gland with absence of the pituitary shaft.

2 DISCUSSION

As mentioned above, Pickardt-Fahlbusch syndrome is defined by the triad: thin or interrupted pituitary nail, adenohypophyseal aplasia or hypoplasia, and absence or ectopia of the neurohypophysis. This pathology most often affects male patients and its incidence has increased due to technological advances in diagnostic methods, such as magnetic resonance imaging and radiology as a whole, which has improved the prognosis of the disease, since it allows an early management with the proper hormonal therapy (BOLINELLI et al, 2021).

The referred syndrome, despite the increase in incidence, is rare, congenital, and its clinical picture is composed of symptoms of tertiary hypothyroidism, that is, that caused by hypothalamic deficiency in the production of HRT and this is caused by the interruption of the portal veins between the hypothalamus and the adenohypophysis (GUTCH et al, 2014). The signs and symptoms are related to low hormone rates, which can generate panhypopituitarism. The clinic presents itself in the patient in the various phases of his life and in childhood causes growth retardation, short stature, intellectual delay, hypotension and seizures. In adolescence, the main clinical point is late puberty, that is, after 13 years in girls and after 14 years in boys (GOSI; KANDURI; GARLA, 2019).

As previously pointed out, the diagnosis is made from the imaging of the pituitary and hypothalamus with magnetic resonance imaging, in which you will observe the ectopic or absent neurohypophysis, the absent or interrupted pituitary stem and a tiny adenohypophysis (BOLINELLI et al, 2021).

Finally, the prognosis is varied and depends on several factors, the main one being the speed of diagnosis and the early treatment of hormonal changes (BAR et al, 2015). In addition, children with Pickardt-Fahlbusch syndrome should be followed during their development and any intellectual delays, growth retardation and/or other symptoms should be addressed individually according to what best benefits the patient at the time, looking for adjacent causes and treating them. In addition, other etiologies, however rare, should always be thought of as differential diagnoses, because if diagnosed, early treatment benefits the patient and generates a good prognosis for the patient (ALALI; SAAD; KABALAN, 2020).

3 CONCLUSION

Pickardt syndrome is a variation of pituitary stem disruption syndrome, which consists of a rare congenital syndrome described by tertiary hypothyroidism caused by disruption of blood flow from the portal vessels connecting the hypothalamus and adenohypophysis. Other typical features besides the classic tertiary hypothyroidism with low thyroid-stimulating hormone are high prolactin, short stature, growth rate retardation, delayed puberty and altered metabolism (GUTCH et al., 2014).

An explanation for the descriptions of this congenital anomaly is that, due to the interruption of blood flow from the portal vessels, there is a blockage of the thyrotropin-releasing hormone (TRH),

endogenous reach the cells in the anterior region of the pituitary gland and consequently the release of TSH is very low, leading to a low stimulation of the thyroid with consecutive hypothyroidism (PICKARDT et al, 1972; GUYTON et al., 2017).

The identification of the syndrome and its early treatment influence both prognosis and quality of life in patients with hypopituitarism and hypothyroidism (CHANDIO et al., 2020). Thus, the deficiency of the action of the pituitary hormone leads to morbidities over the years.

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