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CASE REPORT

Airway approach in a pediatric patient with Noonan syndrome: Presentation of a case

Abordaje de la vía aérea en paciente pediátrico con síndrome de Noonan. Presentación de un caso

Abordagem das vias aéreas em paciente pediátrico com síndrome de Noonan. Apresentação de um caso

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ABSTRACT

Noonan syndrome is a congenital of particular interest to the anesthesiologist because it is considered a probable difficult airway. A 13-year-old male patient was presented with a personal pathological history of Noonan syndrome genotype PTPN11, hypothyroidism, mild intellectual disability who attended the Surgical Guard Corps due to abdominal pain of 16 hours duration. He was diagnosed with acute appendicitis and was scheduled for emergency appendectomy. The presence of Noonan syndrome in the patient required a personalized anesthetic approach to manage potential intraoperative complications. Meticulous preoperative planning, the choice of rapid sequence for anesthetic induction, and rigorous monitoring were key to the success of the surgery.

Keywords: anesthesia; congenital defect; genetic disease; intubation; rasopathy; Noonan syndrome; airway; difficult airway

RESUMEN

El síndrome de Noonan es una enfermedad congénita, de particular interés para el anestesiólogo por ser considerado probable vía aérea difícil.Se presentó un paciente masculino de 13 años con antecedentes patológicos personales de síndrome de Noonan genotipo PTPN11, hipotiroidismo, discapacidad intelectual ligeraque acudió a Cuerpo de Guardia de Cirugía por presentar dolor abdominal de 16 horas de evolución. Fue diagnosticado con apendicitis aguda y se anunció para apendicectomía de urgencia.La presencia del síndrome de Noonan en el paciente requirió un enfoque anestésico personalizado para manejar las posibles complicaciones transoperatorias. La planificación preoperatoria meticulosa, la elección de la secuencia rápida para la inducción anestésica y la monitorización rigurosa fueron clave para el éxito de la cirugía.

Palabas clave:anestesia; defecto congénito; enfermedad genética; intubación; rasopatía; síndrome de Noonan; vía aérea; vía aérea difícil

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RESUMO

A síndrome de Noonan é uma doença congênita causada por uma mutação autossômica dominante. Essa síndrome é de particular interesse para o anestesiologista por ser considerada uma provável via aérea difícil. Paciente do sexo masculino, 13 anos, apresentou história patológica pessoal de síndrome de Noonan, genótipo PTPN11, hipotireoidismo e deficiência intelectual leve, que compareceu ao Corpo de Guardas Cirúrgicos devido a dor abdominal com duração de 16 horas. Ele foi diagnosticado com apendicite aguda e foi agendado para apendicectomia de emergência. A presença da síndrome de Noonan no paciente exigiu abordagem anestésica personalizada para manejo de possíveis complicações intraoperatórias. O planejamento pré-operatório meticuloso, a escolha da sequência rápida para indução anestésica e o acompanhamento rigoroso foram fundamentais para o sucesso da cirurgia.

Palavras-chave: anestesia; defeito congênito; doença genética; intubação; rasopatia; Síndrome de Noonan; via aérea; via aérea difícil

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INTRODUCTION

RASopathies are a group of congenital defects caused by mutations in genes encoding components or regulators of the RAS/MAPK pathway. They represent one of the most prevalent groups of genetic diseases, including: Noonan syndrome, Noonan-like with anagen hair, Noonan-like with or without elevated risk of juvenile myelomonocytic leukemia, Noonan with multiple lentigines, Cardiofaciocutaneous, Costello, Neurofibromatosis-Noonan and Legius.(1)

Noonan syndrome (NS) is a congenital disease caused by an autosomal dominant mutation. It was first described by Noonan and Ehmke in 1963, the expression of this condition varies according to age and it behaves clinically as a multisystem disorder.(2) It is characterized by a peculiar phenotypic triad: craniofacial anomalies giving a typical facies, congenital heart disease and short stature, in addition to varying degrees of intellectual disability.(3) It has an incidence of 1:1000 to 1:1000 in the first two years of life.

It has an incidence of 1:1000 to 1:2500 live births, with equal prevalence between genders. SN owes its origin to a mutated gene, being more frequent the mutation of the PTPN11 gene, which predominates in approximately 50% of patients; the SOS1, KRAS, NRAS, SHOC2, CBL and RAF1 genes, together, cause approximately 80% of clinically diagnosed cases of SN.(4)

The main signs found in this syndrome are palpebral ptosis, hypertelorism, downward slanting palpebral fissures, low implantation of auricular pavilions with posterior rotation and thickened helix, short neck with excess skin on the nape of the neck and low posterior hairline, rib cage deformity and cryptorchidism.(5)

The most frequent symptoms that may occur are lymphatic dysplasia, coagulation disorders and feeding problems during infancy. Among the cardiological alterations, the most common are pulmonary valve stenosis and hypertrophic cardiomyopathy.[6] At the cutaneous level, its manifestations will depend on the causal mutation and café-au-lait spots, pigmented nevus and lentigines may be observed.[7]

This syndrome is of particular interest to the anesthesiologist, as it is considered likely to have a difficult airway, short stature, facial dysmorphia, hemostasis, musculoskeletal and cardiological alterations, whose evolution conditions life expectancy, reaching advanced ages nowadays thanks to their control.[8] That is why this article aims to present the case of a patient with NS with a difficult airway.

CASE PRESENTATION

A 13-year-old male patient with a personal pathological history of SN genotype PTPN11, hypothyroidism and mild intellectual disability and family pathological history of maternal hypothyroidism and arterial hypertension in several family members. He is being treated with growth hormone (somatotropin 0.6ng/day) and Levothyroxine sodium 0.1mg/day. He goes to the surgical ward for presenting abdominal pain of 16 hours of evolution, initially periumbilical that later moved to the right iliac fossa, febrile and nausea that does not lead to vomiting, fasting for approximately 8 hours.

Physical examination

Fascia: broad forehead, separated eyebrows, antimongoloid eyes, broad-based nose, micrognathia, low-set ears. Chest: upper pectuscarinatum and lower pectusexcavatum, wide mammillary line with widely separated nipples.

Figure 1 Photograph of the patient showing facial and thoracic deformities.

Respiratory auscultation without alterations: _ Cardiovascular auscultation: systolic murmur II/VI at the base. Vital signs: Blood pressure: 110/70 mmHg Heart rate: 78 per minute Temperature: 36.8 ° C Partial oxygen saturation: 100%. Central nervous system: Responds to simple verbal commands, Conscious and oriented in time, space and person.

Complementary tests
Hemoglobin: 117g/dL
Hematocrit: 35%.
Coagulation: Bleeding Time: 1 minute Clotting Time: 7 minutes Platelet Count: 380x10(9/L) - Prothrombin Time: 14 seconds - Activated Partial Thromboplastin Time: 34 seconds - Blood Group: B positive
Leukocytes: 11x10(g/dL) Polymorphic nuclear: 60% Lymphocytes: 39.3% Eosinophils 001%.

Imaging studies: Posteroanterior Chest X-ray at telecardiogramdistance: Variable bone density,Cervico-dorsal scoliosis, horizontal ribs, subglottic tracheal stenosis of approximately 5 cm in length and thickness less than a third of the rest of the tracheal silhouette. Cardiothoracic index slightly increased at the expense of left cavities, widening of the superior mediastinum,bilateralbronchovascular enhancement.

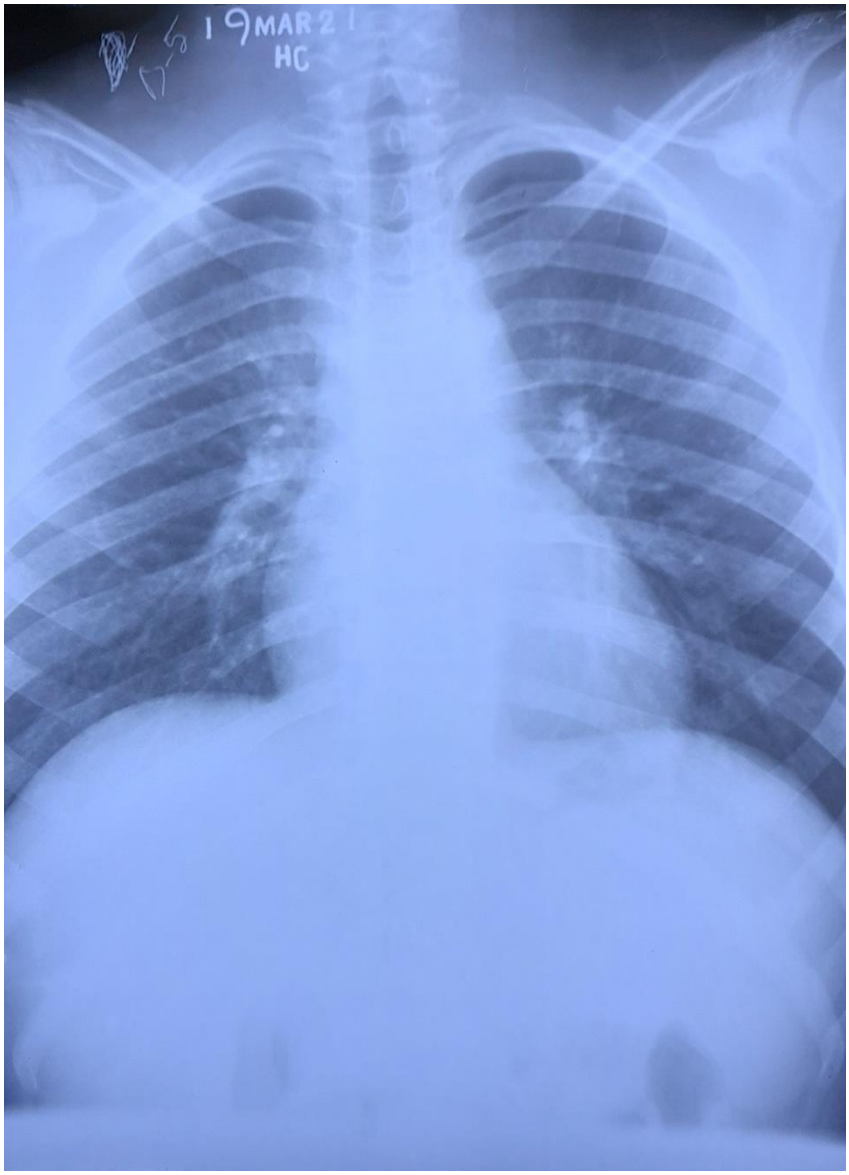

Figura 2. Radiografía de tórax vista postero-anterior.
Figure 2 Chest X-ray postero-anterior view.

Acute appendicitis was diagnosed and emergency appendectomy was announced, after evaluation with the Cardiology and Endocrinology specialties and communication with the specialist of Medical Genetics via videocall, confirming the presence of mild stenosis of the pulmonary semilunar valve and follow-up in consultation at the Center for Genetic Engineering and Biotechnology.

Anesthetic Management: Preoperative: Hydration: Volume replacement by fasting with 0.9% saline solution. Monitoring: Continuous electrocardiogram in lead II, heart rate, pulse oximetry; temperature, end-expiratory carbon dioxide, spontaneous diuresis measurement Premedication: Desmopressin acetate 20 minutes prior to anesthetic induction. Availability of volumetric expanders and compatible blood products was guaranteed. Anesthetic induction: Rapid sequence with propofol 3.5mg/kg and rocuronium 0.6mg/kg with ventilatory assistance by face mask coupled to a semi-open Jackson-Rees circuit. Effective oro-tracheal intubation at the first attempt with oro-tracheal cannula No. 5 with cuff, Maintenance: Total Intravenous Anesthesia with propofol 4mg/kg/h, fentanyl 5ug/kg/h rocuronium 0.6mg/kg/h, controlled ventilation with intermittent positive pressure ventilation mode, total volume 7ml/kg, respiratory rate 14 per minute, inhaled O2+ nitrous oxide 50%, maintaining hemodynamic stability.

Surgical time: 45 minutes. McBurney incision appendectomy was performed, confirming acute catarrhal appendicitis with blood loss of less than 100ml. No surgical, anesthetic or hemodynamic complications. Volumetric replacement according to trans-surgical losses with crystalloids (Ringer Lactate 200ml)

Recovery: Spontaneous reversal of neuromuscular blockade and satisfactory recovery of the level of wakefulness. Extubation with the patient awake after aspiration of secretions

Postoperative analgesia: Sodium Metamizole 1g intravenous infusion diluted in 200ml NaCl 0.9%. The patient maintains clinical stability, recovering satisfactorily.

DISCUSSION

The anesthetic management of NS is a challenge due to its particularities; in the literature, the anesthetic technique in patients with this condition is described as a complex procedure due to its musculoskeletal malformations at the lumbar level and the incomplete motor and sensory block.[9]

The anesthetic approach to the adolescent with NS for emergency abdominal surgery tests not only the medical knowledge and skills of the work team but also the physical capacities of the patient to respond to the anesthetic-surgical aggression, since in addition to his basic multi-organ alterations, there are also those of the surgical pathology per se.

A case reported by Moreno, et al.[10] refers that the main facial characteristics in his patient were wide forehead, abundant periorbital tissue, low set ears and posterior rotation, antimongoloid palpebral fissure, palpebral ptosis, depressed nasal bridge, wide nasal tip, flat malar region, prominent cheeks, deep philtrum, data that coincide with the characteristics found in the exposed case, some variations found between both cases may be due to the fact that the patients present mutations in different genes involved in this syndrome.

Martínez,et al.[11] report a case in which their patient on regional physical examination of the thorax only presents a slight pectusexcavatum, which contrasts with the present case, in which the patient shows superior pectuscarinatum and inferior pectusexcavatum, broad mammillary line with widely separated nipples, which could be explained by the age difference between both cases and the phenotypic variations that people with NS suffer throughout their lives.

The authors refer that no reports of other cases of SN with subglottic tracheal stenosis were found in the scientific literature, which makes the present case more striking, since this congenital defect is not characteristic of patients with SN. When subglottic tracheal stenosis is associated with other features of the syndrome such as short neck, upper pectuscarinatum and lower pectusexcavatum, they make the airway approach in this case even more challenging for the anesthesiologist and require more preparation and skill from the anesthesiologist.

The presence of pectusexcavatum, pectuscarinatum and scoliosis are associated bony abnormalities that may limit lung capacity, so look for symptoms of pulmonary dysfunction. The goals in mechanical ventilation are to avoid hypoxia and hypercapnia, situations that generate an increase in pulmonary vascular resistances and right ventricular tension.[12]

FINAL CONSIDERATIONS

The presence of Noonan syndrome in the patient required a personalized anesthetic approach to manage potential transoperative complications. Meticulous preoperative planning, choice of rapid sequence for anesthetic induction, and the rigorous monitoring were the key to the success of the surgery. The present case provides an example that can serve as a reference for specialists faced with a surgical emergency in patients with Noonan syndrome or any other RASopathies.

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Conflicts of interest:
The authors declare that there are no conflicts of interest.

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