

Anaesthesia of an infant with Johanson-Blizzard Syndrome: A case report and review of literature

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ABSTRACT

We present an 8 months old infant with Johanson-Blizzard syndrome (Autosomal recessive disease) for postero sacral anorectal formation for imperforate anus who required general anaesthesia. An anaesthetic plan for each individual should be personalized as the phenotypic presentation varies over a spectrum of severe form to hypomorphic form. The anaesthetic considerations included pancreatic exocrine insufficiency, mental retardation, hearing impairment, growth delay, hypotonia, lax joints and difficult airway due to short nose, absent ala nasi, receding chin, maxillary hypoplasia and high arched palate. Pre-operative sedation was avoided because of hypotonia of the pharyngeal muscles and difficult airway. Pancreatic enzymes replacement continued with intraoperative sugar monitoring. To the best of our knowledge there have been less than hundred cases reported of JB Syndrome, four cases have been reported from India. There are very limited number of reported case describing anaesthetic considerations of JB Syndrome.

Key words: Johanson-Blizzard syndrome–Anaesthetic management–Pancreatic exocrine dysfunction

1 INTRODUCTION

The Johanson-Blizzard (JB) syndrome is a rare syndrome, which is autosomal recessive disease caused by whole exon deletions and duplications in UBR1 gene [1],[2]. It presents with constellation of body system involvement but presence of typical facial features, pancreatic exocrine insufficiency, absent or hypoplastic alae nasi, ectodermal scalp defect, microcephaly, congenital deafness and growth retardation are most common [3]. In India, first case of JB Syndrome was described in 2004 [4]. From India till date four cases have been reported [4],[5],[6], this is first case report discussing about anaesthetic considerations.

2 CASE REPORT

An 8 months old infant, known case of JB Syndrome, presented for postero sacral anorectal formation for imperforate

anus. In family history, one sibling was diagnosed as JB Syndrome, died after 3 months of age and one IUD also at 8 months of gestation was present.

Our patient was full term baby, born by caesarean section from consanguineous marriage with birth weight of 2.8 kg (< 5 percentile), head circumference 31cm (< 5 percentile), body length 44cms (<5 percentile), Apgar score at 1 min was 8 and at 5 min was 9. Genetic testing confirmed UBR1 gene mutation, 2D echocardiography (ECHO) revealed patent foramen ovale with left to right shunt with pulmonary hypertension. He was operated for transverse colostomy in view of imperforate anus which was uneventful. After which, he developed loose stool. Serum amylase and serum lipase were found to be low and pancreatic enzyme replacement therapy started for him.

Out of other traits of JB Syndrome, he had imperforate anus, café au lait spot on buttock and lower limb, absent ala nasi, mongoloid slant, beak shaped nose, frontal upsweep of hair, bilateral simian crease, hypertrichosis and aplasia cutis at anterior and posterior fontanelle.

His current weight was 5.4 kg (< 3 percentile), head circumference 42cm, body length 67 cm (< 3 percentile), de-

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veloped head control at 7 months and was not able to sit or stand with support. Few investigations repeated for him to find out any liver function derangement, anaemia, diabetes, hypothyroidism, hypocalcaemia and cardiac functional status. His haemoglobin was 11.6gm/dl, serum creatinine 0.45mg/dl, random blood sugar 92mg/dl, serum electrolytes, thyroid function tests and 2D ECHO reports were normal. In airway examination, receding chin, no teeth, maxillary hypoplasia, high arched palate and absent ala nasi were present. Hypotonia was present but there was no history of any respiratory compromise or regurgitation. Anaesthesia was planned by general anaesthesia with endotracheal intubation of trachea and controlled ventilation. No premedication was given in view of anticipated difficult airway. Operation Theatre was prepared for special paediatric case with difficult airway cart. The patient was induced with inhalational agent Sevoflurane 4% with oxygen and intravenous cannulation done in right upper and lower limb by 24G, Vasofix, BBraun. Injection (inj) Fentanyl was given in dose of 1microgm/kg and bag and mask ventilation was checked which was found to be adequate. Then inj Atracurium was given in dose of 0.5mg/kg, and laryngoscopy performed with straight blade Magill forcep size 0. In two attempts uncuffed entotracheal tube of size 4 secured and confirmed for position. Mechanical ventilation started with pressure controlled ventilation. Caudal block was performed in lateral position and 3ml 0.25% Bupivacaine given and Inj Paracetamol 100mg given intravenously. Proper padding done for lax joints and prone position. Maintenance of anaesthesia was done by oxygen, air and Isoflurane. Intra operatively temperature, urine output, neuromuscular monitoring and blood sugar monitoring was done, which remained in range of 83 – 96mg/dl. Patient remained hemodynamically stable throughout the surgery. At the end of the surgery reversal of anaesthesia was done by Inj Atropine and Inj Neostigmine. The trachea was extubated and all reflexes regained. The patient remained pain free in post-operative period and monitored extensively for 2 days in HDU for any respiratory compromise.

3 DISCUSSION

The anaesthesiologist must consider several factors in management of JB Syndrome as it presents with constellation of body system involvement, out of which pancreatic exocrine insufficiency, abnormal hair pattern, alopecia, failure to thrive, intrauterine growth retardation, malabsorption, short nose, short stature and underdeveloped nasal alae are most common one [1]. Figure 1 shows characteristic facial features and Table 1 enlists all the associated conditions of anaesthetic importance.

Careful preoperative evaluation and optimization is must especially pancreatic exocrine functions as it affects 80-99% [1] patients and makes them prone for diarrhoea and electrolyte imbalances. Endocrine functions can be affected and may lead to diabetes [1], hypothyroidism [1] and hypopituitarism [10]. Hepatic failure [1] and cardiac manifestations

Table 1. (Original) Features of Johanson-Blizzard syndrome of anaesthetic considerations

Serial number	Feature
1.	Exocrine pancreatic insufficiency [1]
2.	Intrauterine growth retardation [1], short for gestational age
3.	Intellectual disability [1], sensorineural hearing loss [1] (difficult communication)
4.	Diabetes mellitus [1], hypothyroidism [1], hypocalcemia [1]
5.	Hepatic failure [1], Cholestasis [7], Malabsorption, hypoproteinemia, edema (hepatic involvement)
6.	Short nose [1], underdeveloped alae nasi [1] , maxillary hypoplasia, microdontia, oligodontia, microcephaly [8] , high arched palate (difficult airway)
7.	Anaemia [3] , Mild to moderate thrombocytopenia [3]
8.	Dilated cardiomyopathy [1], atrial septal defect [1], ventricular septal defect [1], situs inversus totalis (cardiac involvement)
9.	Joint laxity [1] (proper positioning)
10.	Hydronephrosis [9] (renal involvement)
11.	Muscular hypotonia [7] (careful use of neuromuscular blocking agents)



Figure 1. Infant showing characteristic features with abnormal hair line, absent ala nasi and short nose

[1] like dilated cardiomyopathy, atrial and ventricular septal defect are not very common, but if present then can make anaesthesia more challenging. Other not very common manifestation noted in patients of JB Syndrome is hypocalcemia [1] which should also be ruled out and properly optimized. So proper drug history and adjust of anaesthetic drug doses based on hepatic enzyme functions are crucial.

Absent or underdeveloped ala nasi, short nose, receding chin, small and abnormal teeth, maxillary hypoplasia and elevated palate can make bag and mask ventilation and in-

tubation difficult, which was present in our case. Regurgitation can also occur in patients with hypotonia, and mandated titrated use of neuromuscular blocking agents. Difficult airway cart should be kept ready. In our patient bag and mask ventilation was optimal but endotracheal intubation was little difficult due to high arched palate.

They may present as premature and small for gestational age^[1], which has its own concerns. As older kids, deafness and mental retardation can make the communication difficult. Overall knowing the patient well and optimizing associated conditions should be key of uneventful and successful management.

4 CONCLUSIONS

Anaesthetic management of patients with JB Syndrome should be individualised because of multisystem involvement and varying pattern among patients. Difficult airway, pancreatic function optimisation, cardiac involvement and mental retardation are important challenges. [1–10]

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