

BECKWITH-WIEDEMANN SYNDROME WITH CLEFT PALATE

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BECKWITH-WIEDEMANN SYNDROME WITH CLEFT PALATE (ABSTRACT): Beckwith-Wiedemann syndrome is a rare congenital disorder. Early diagnosis, proper counselling of parents is essential because of significant risk of subsequent development of malignant tumours, associated with this syndrome. Association of cleft palate in this syndrome is extremely rare. Specific difficulties and risks posed by the craniofacial abnormalities like macroglossia and cleft palate are highlighted and discussed along with the management options. The literature is reviewed and discussed.

KEY WORDS: BECKWITH-WIEDEMANN SYNDROME, CLEFT PALATE, OMPHALOCELE

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INTRODUCTION

Beckwith Wiedemann Syndrome (BWS) is a rare congenital disease of low prevalence independently recognised by Beckwith in 1963 and Wiedemann in 1964. Association of this syndrome with cleft palate is even rarer. Surgical corrections of the craniofacial abnormalities pose significant and specific challenges.

CASE REPORT

A term, large for gestational age female baby was born to 29 years old primi, which had undergone antenatal checkups outside and was referred to this hospital for non progress of labour, through Caesarian section on 04/09/2007. The baby weighed 4500 grams (>95th centile), with head circumference of 36 cm. (>75th centile) and length of 55 cm. (>95th centile). The APGAR score was 5 at 1 minute and 10 at 5 minutes. The notable congenital anomalies were omphalocele with intact sac, macroglossia with protruding tongue (Fig. 1), short neck and linear indentations of ear lobule with posterior helical pits (Fig. 2), nevus flammeus and cleft palate (Fig. 3). The baby had an episode of generalized tonic clonic convulsion; blood sugar estimated at that time was 45 mg%. Euglycemia was attained and subsequently maintained by intravenous dextrose infusion. A clinical diagnosis of Beckwith-Wiedemann syndrome was made on the basis of these clinical features. Omphalocele was dressed with sterile vaseline gauze and pads and baby started on parenteral antibiotics, put on IV fluid and nasogastric decompression. The baby passed meconium within 24 hours of birth.

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Subsequent investigations included haematology, renal function tests, serum electrolytes including calcium, liver function tests, CRP, blood culture and all were within normal limits. Ultrasonography of whole abdomen, CT scan of cranium and brain and echocardiography did not reveal any abnormality.



Fig. 1 *Omphalocele with macroglossia*



Fig. 2 *Indentations of ear lobule with posterior helical pits*



Fig. 3 *Cleft palate*

Parents did not have any feature of the syndrome. Both the parents were counselled and educated about the syndrome, regarding hypoglycaemia with possible risks of neurological consequences and its prevention, subsequent risk of tumours of solid organs. They were also counselled regarding the small risk during the subsequent pregnancies.

The child underwent a successful primary fascial repair of the omphalocele under general anaesthesia with endotracheal intubation and intraoperative plasma glucose monitoring, two days after admission.

The post operative period was uneventful and was discharged on the 20th post operative day once parents were confident about the feeding techniques for the child.

The child underwent repair of the cleft palate and release of ankyloglossia under general anaesthesia at the age of 11 months. The follow up of the child till age of two and half years with serial ultrasound examinations of the abdomen did not reveal any evidence of solid tumours. The child has difficulty in articulation and is under speech therapy.

DISCUSSION

Beckwith-Wiedemann syndrome is a clinical diagnosis, criteria for diagnosis being presence of three major findings (macroglossia, pre- or post natal growth greater than 90th percentile and abdominal wall defects) or two major plus minor manifestations (ear creases or pits, facial neavus flammeus, hypoglycemia, nephromegaly, hemihypertrophy) [1]. Association of cleft palate as a part of the syndrome is extremely rare and very few cases have been reported [2,3]. The genetics of this syndrome is complex, imprinted genes of chromosome 11p15 have been implicated in both familial and sporadic varieties. Paternally derived duplications of chromosome 11p15 and maternally inherited inversions or balanced translocations may be associated this syndrome [4].

The children with this syndrome have increased risk of developing subsequent neoplasia like Wilm's tumour, hepatoblastoma, neuroblastoma, rhabdomyosarcoma, adrenocortical carcinoma, estimated to be around 7.5% in the first eight years of life, rarely after 10 years of age [5].

Airway management with craniofacial abnormalities like macroglossia compounded by presence of cleft palate in Beckwith-Weidemann syndrome poses serious challenges. The normal bony and soft tissue anatomy is altered; the anaesthesiologist must be aware and familiar with it [6]. Difficulty in bag/mask ventilation and endotracheal intubation following the induction of anaesthesia and muscle paralysis is to be anticipated, so preparations for a difficult airway management need to be considered before induction [7].

Surgical correction of cleft palate in a patient of Beckwith-Weidemann syndrome is advisable after the age of six months before the speech development process starts [2]. The enlarged tongue has both functional and cosmetic deformity, which may affect the oral airway, speech, and the development of the jaws.

Tongue reduction surgery is advocated, preferably to precede the cleft palate repair or may be combined with the palate surgery to reduce the repeated anaesthetic procedural risk. Articulation errors have been reported by various investigators due to the craniofacial abnormalities in Beckwith-Weidemann syndrome. However data are limited as to define the nature of the articulation problems. In a study of 40 patients of Beckwith-Weidemann syndrome with macroglossia, 29 of them had articulation errors, and it persisted in some after corrective surgery [8]. A phonetic analysis of patients of Beckwith-Wiedemann syndrome with macroglossia revealed the affection of consonants with an anterior place of articulation, related to inappropriate tongue and lip postures. An observer experiment conducted, in which naive and expert observers rated speech samples from three modes of presentation (auditory-only, visual-only, and audiovisual), showed that the subjects' speech was more disturbed visually than auditorily [9].

Phonetic and articulation errors are also known to occur after a cleft palate repair. Hypernasality, weak pressure consonants, nasal escape may indicate incompetent nasopharyngeal sphincter.

Dental malocclusion with repaired clefts involving the alveolar ridge will distort the sounds „s z ch j, sh, zh”. Good improvement is usually noticeable after about three months of regular weekly sessions of speech therapy with a young child of average intelligence [10]. The value of tongue volume reduction surgery for improvement of speech needs prospective assessment and that the adequacy of direct articulation therapy in Beckwith-Wiedemann Syndrome needs to be evaluated [9]. It is also suggested that social environment can also be a contributing factor for development of phonation and articulation [2].

CONCLUSIONS

Beckwith-Wiedemann syndrome is a rare congenital disorder. Early diagnosis is essential because of the significant risk of subsequent development of malignant tumours, associated with this syndrome. Association of cleft palate in this syndrome is extremely rare. The treatment of patients with cleft palate and Beckwith-Wiedemann syndrome are difficult and represents a challenge for surgeon.

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