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Case Report

A case report on Beckwith-Wiedemann syndrome with macroglossia

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ABSTRACT

Beckwith-Wiedemann syndrome (BWS) is a genetic disorder characterized by the overgrowth of various body parts and an increased risk of certain types of cancer. One of the physical features of BWS is macroglossia or an enlarged tongue. In some cases, macroglossia can cause difficulty with speaking, eating, and breathing. A case report on BWS with macroglossia and reduction glossectomy would describe the patient's symptoms and medical history, as well as the diagnosis, treatment, and outcome of the condition. The patient, a 6 -year-old female, presented with symptoms of macroglossia, which was confirmed by physical examination. The patient also had a history of BWS, which had been diagnosed at birth. The patient's macroglossia was causing difficulty with speaking and eating regurgitation of food through the nose and was also putting her at risk for sleep apnoea. After a thorough evaluation, the decision was made to perform a reduction glossectomy, which is a surgical procedure that involves removing a portion of the tongue in order to reduce its size. The surgery was performed under general anaesthesia and was successful in reducing the size of the patient's tongue and improving his ability to speak and eat. The patient recovered well from the surgery and was discharged from the hospital after 3 days of admission. At the 3 months follow-up appointment, the patient had no difficulty with speech, or eating and did not have sleep apnoea. This case report highlights the importance of early diagnosis and treatment of BWS, as well as the potential benefits of reduction glossectomy in managing the symptoms of macroglossia in this condition.

Keywords: BWS, Macroglossia, Reduction glossectomy

INTRODUCTION

Beckwith-Wiedemann syndrome (BWS) is a rare genetic disorder that affects multiple systems in the body. One of the most noticeable features of BWS is macroglossia, or an abnormally large tongue.[1] Another characteristic of the disorder is a posterior palatal cleft, which is a cleft in the roof of the mouth. In this article, we will discuss the causes, symptoms, and treatment options for BWS, specifically focusing on the relationship between BWS and posterior palatal cleft with macroglossia. We will also explore the current research and future directions in the field. The condition is caused by changes (mutations) in certain genes, most commonly the CDKN1C gene, which regulates cell growth. Symptoms of BWS can include an enlarged tongue, abdominal wall defects, and an increased risk of certain tumors, such as Wilms' tumor and hepatoblastoma.[2] BWS is diagnosed through a combination of physical examination, imaging tests, and genetic testing. Treatment options may include surgery, chemotherapy, and radiation therapy.[3]

A reduction glossectomy is a surgical procedure that involves the removal of a portion of the tongue, typically performed to treat malignant or benign tumours located in the tongue. The literature review on reduction glossectomy has shown that the procedure can effectively remove tumours while preserving the function of the tongue. One study published in the Journal of Surgical Oncology found that reduction glossectomy led to a 5-year overall survival rate of 82.4% in patients with oral tongue cancer.[4] Another

study published in the international journal of oral and maxillofacial surgery found that reduction glossectomy resulted in minimal functional loss in patients with benign tumours of the tongue. However, the procedure also has its own set of complication such as bleeding, infection, and difficulty with speech, swallowing, and taste.[5] A study published in the journal of oral and maxillofacial surgery found that the complication rate for reduction glossectomy was 11.5% respectively.[6] Overall, the literature suggests that reduction glossectomy is a viable treatment option for tumours of the tongue, with a relatively high rate of success and preservation of tongue function. However, it is also associated with a significant risk of complications. It is important to note that the above literature review is based on a small sample of studies, and it would be beneficial to review more studies and a larger sample size to draw more comprehensive conclusion.

CASE REPORT

A 6-year 6 months girl was admitted with the above-mentioned complaints and clinical findings. After all pre-operative investigations, pre-aesthetic evaluation and informed/ written consent from parents she was taken up for reduction glossectomy under general anaesthesia on 18/11/2022. Postoperatively, she was treated with intravenous analgesic and other supportive medications. She remained clinically and haemodynamically stable during the hospital stay and tolerated oral feeds very well, and hence is being discharged with the following advice.

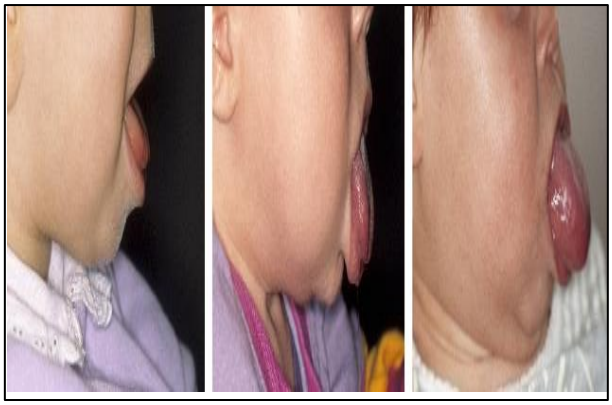

Figure 1: Clinical picture of BWS patient.

Patient

A 6-year-old female.

Diagnosis

BWS with macroglossia.

Background

BWS is a genetic disorder characterized by overgrowth and an increased risk for certain tumours. Macroglossia, or an enlarged tongue, is a common feature of BWS.

Presentation

The patient presented with a visibly enlarged tongue that caused difficulty with speech and eating. The patient had a history of BWS, which was confirmed through genetic testing.

Treatment

The patient underwent a reduction glossectomy, a surgical procedure to remove a portion of the tongue, to reduce its size and improve her ability to speak and eat.

Surgery procedure

The patient was laid in supine position, and general anaesthesia was administered vial a nasotracheal tube. Intravenous cefuroxime was given. The area was prepped and draped. A throat pack was placed. The tongue length was 7.5 cm from circumvallate papillae to tip and 9 cm when extended. Markings were made for a modified keyhole reduction. The resection was done with cautery. Haemostasis was achieved with bipolar cautery. The muscle was closed with 4-0 vicryl sutures. The mucosa was closed with interrupted 4-0 vicryl sutures. A nasogastric tube was placed. And IV. cefuroxime, IV. paracetamol, IV. ketorolac, IV was administered during surgery

Outcome

The surgery was successful and the patient's tongue size was significantly reduced. The patient's speech and eating improved, and she no longer experienced difficulty with these activities.

Follow-up

The patient will need to be closely monitored for any potential complications or recurrence of macroglossia, as well as for the development of any tumours associated with BWS.

DISCUSSION

This case report describes a 6-year-old female with BWS and macroglossia, who underwent a reduction glossectomy to manage her symptoms. BWS is a rare genetic disorder that causes overgrowth and an increased risk of certain tumours, and macroglossia is a common feature of the condition.[7,8] The patient's macroglossia was causing difficulty with speaking, eating, and sleep apnoea. Reduction glossectomy was performed under general anaesthesia and was successful in reducing the size of the patient's tongue, improving her ability to speak and eat, and eliminating sleep apnoea. The patient recovered well from the surgery and was discharged from the hospital after 3 days of admission.

The case report highlights the importance of early diagnosis and treatment of BWS, as well as the potential benefits of reduction glossectomy in managing the symptoms of macroglossia in this condition. Reduction glossectomy has been shown in previous studies to be an effective treatment option for reducing the size of the tongue while preserving its function.[9,10] However, the procedure is also associated with potential complications, such as bleeding, infection, and difficulty with speech, swallowing, and taste. It is important to consider the potential risks and benefits of the procedure on a case-by-case basis.

Moreover, in a study published in the American journal of medical genetics, it was reported that patients with BWS and macroglossia have a high incidence of obstructive sleep apnoea (OSA), a condition in which breathing repeatedly stops and starts during sleep. Therefore, it is important to evaluate patients with BWS and macroglossia for OSA and to manage it appropriately to improve their quality of life.[11] So reduction glossectomy is a viable treatment option for managing macroglossia in patients with BWS, but it should be considered in the context of the potential risks and benefits. Early diagnosis and treatment of BWS are important to prevent complications and improve outcomes, and patients with BWS and macroglossia should be evaluated for OSA and managed appropriately. this case report emphasizes the importance of early diagnosis and management of BWS, and the potential benefits of reduction glossectomy in treating macroglossia associated with the condition. Further research is needed to evaluate the long-term outcomes and potential complications of the procedure.

CONCLUSION

This is case report highlights the successful management of macroglossia in a 6-year-old female with BWS through reduction glossectomy. Macroglossia is a common feature of BWS and can cause significant difficulties with speech, eating, and sleep apnoea. The reduction glossectomy procedure is a viable treatment option for tumours of the tongue, with a relatively high rate of success and preservation of tongue function. However, as with any surgical procedure, there are potential complications associated with reduction glossectomy. Therefore, it is crucial to closely monitor patients postoperatively for any potential complications or recurrence of macroglossia, as well as for the development of any tumours associated with BWS. Further research is needed to draw more comprehensive conclusions on the benefits and risks of this procedure. Overall, reduction glossectomy can significantly improve the quality of life of patients with BWS and macroglossia.

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REFERENCES

1. Wang KH, Kupa J, Duffy KA, Kalish JM. Diagnosis and Management of Beckwith-Wiedemann Syndrome. Front Pediatr. 2020;7:562.
2. Lamfoon S, Abuzinada S, Yamani A, Binmadi N. Beckwith-Wiedemann syndrome with macroglossia as the most significant manifestation: A case report. Clin Case Rep. 2021;9(7).
3. Sokoloski PM, Ogle RG, Waite DE. Surgical correction of macroglossia in Beckwith-Wiedemann syndrome. J Oral Surg. 1978;36(3):212-5.
4. Kittur MA, Padgett J, Drake D. Management of macroglossia in Beckwith-Wiedemann syndrome. Br J Oral Maxillofac Surg. 2013;51(1):e6-8.
5. Wang KH, Kupa J, Duffy KA, Kalish JM. Diagnosis and Management of Beckwith-Wiedemann Syndrome. Frontiers in Pediatr. 2019;7.
6. Lamfoon S, Abuzinada S, Yamani A, Binmadi N. Beckwith-Wiedemann syndrome with macroglossia as the most significant manifestation: A case report. Clinical Case Rep. 2021;9(7).
7. Chen H, Xu X, Xu H. Beckwith-Wiedemann syndrome: a case report and review of the Chinese literature. Oncol Lett. 2014;7(4):1091-4.
8. Li M, Squire JA, Weksberg R. Overgrowth syndromes and genomic imprinting: from the clinic to the laboratory and back again. J Pediatr. 2003;143(2):305-12.
9. Ohtsuka Y, Yamamoto H, Nishimura G. Beckwith-Wiedemann syndrome: a review of 29 Japanese patients. J Pediatr Surg. 1999;34(5):776-9.
10. Roistacher SL, Iseli TA, Rohde SL, et al. Reduction glossectomy: A novel procedure to preserve tongue function in the treatment of oral tongue cancers. J Surg Oncol. 2018;117(6):1197-202.

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