CASE REPORT

Oral polyp as the presenting feature of Beckwith-Wiedemann syndrome in a child

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SUMMARY

Beckwith-Wiedemann syndrome (BWS) is a congenital growth disorder characterised by abdominal wall defects, macroglossia and somatic gigantism. A number of associated features, including gastrointestinal and urinary tract polyps, have been described, but there are no previous reports of oral polyps occurring in this syndrome. We describe the first case of BWS presenting with an oral polyp. Clinicians should be alert to the possibility of BWS if other features of the syndrome are present, in children with oral polyps.

BACKGROUND

Beckwith-Wiedemann syndrome (BWS) is a congenital disorder characterised typically by abdominal wall defects, macroglossia, somatic gigantism, hemihypertrophy and neonatal hypoglycaemia. A number of associated features have been described including naevus flammeus, exophthalmos, neonatal hypoglycaemia and cardiac abnormalities.¹ Of particular importance is that children with BWS are at a much higher risk of embryonic tumours including rhabdomyosarcoma, neuroblastoma and nephroblastoma. Various surveillance protocols have therefore been recommended to attempt to reduce morbidity and mortality from malignancy in these children.²

BWS is a rare disorder with an estimated incidence of 1 of 13 700.¹ A variety of genetic or epigenetic alterations in growth regulatory genes on chromosome 11p15.5 are associated with specific phenotypes. BWS is usually sporadic but familial transmission is estimated to occur in around 15% of cases.¹

While gastrointestinal (GI)³ and urethral polyps⁴ have both previously been reported in BWS, an association with oral polyps has not previously been described. We present the case of a 4-year-old girl with a large oral polyp, who was subsequently diagnosed with BWS. Clinicians should be alert to the possibility of BWS in children with oral polyps if other signs or symptoms of the syndrome are present.

CASE PRESENTATION

A 4-year-old Saudi girl presented to the oral medicine outpatient clinic with her family. She had a 4-week history of poor oral intake and difficulty in swallowing. There was marked developmental delay in speech and intelligence. The patient had congenital hypothyroidism and took oral thyroxine. On clinical examination, her weight was 18.3 kg (75th–91st centile) and her height was 112 cm

(98th centile). Her face appeared asymmetrical with right-sided hyperplasia. The ears and earlobes were an abnormal shape. Examination of the oral cavity demonstrated macroglossia with depapillation of the tongue. On palpation, the tongue was smooth, non-tender and slightly larger on the right. A well-defined solitary pedunculated growth was seen on the right buccal mucosa opposite to the first primary lower molar, measuring 8 mm in maximum diameter. The growth was non-tender, smooth and soft in consistency, with the surrounding areas showing no erythema or induration. A surgical excision was performed at a later date. The speech delay and difficulty in swallowing was attributed to the macroglossia, which may have been compounded by the polyp.

INVESTIGATIONS

An upper GI endoscopy was performed to rule out obstructive causes of dysphagia, and was normal. Histopathology assessment of the polyp demonstrated hypertrophic skeletal muscle cells with evidence of vacuolation (figure 1A, B). Further immunohistochemical staining using myoglobin antibodies was undertaken to confirm the polyp was of skeletal muscle origin (figure 2). A panel of immunohistochemical markers were also used (table 1).

In view of the facial asymmetry, congenital hypothyroidism and abnormal earlobes, a working diagnosis of BWS was made. The patient was referred onwards for genetic testing. Genetic analysis showed a mutation in the gene CDKN1C, confirming the clinical diagnosis of BWS.

DIFFERENTIAL DIAGNOSIS

The differential diagnosis of oral polyps in children include skeletal muscle tumours such as rhabdomyomas⁵ or rhabdomyosarcomas,⁶ granular cell tumours,⁷ paragangliomas⁸ and benign lipomatous tumours⁹ such as lipomas, lipoblastomas and hibernomas.

TREATMENT

Surgical excision was performed as the treatment of choice.

OUTCOME AND FOLLOW-UP

The patient made an unremarkable recovery. There was no reoccurrence at 6-month follow-up

DISCUSSION

BWS, while rare, is still the most common genetic overgrowth syndrome. It is characterised by prenatal

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Figure 1 Microscopic photographs of the examined specimen (H&E, (A) (×100), (B) (×200)).

and postnatal overgrowth with a number of other features including polyhydramnios, abdominal wall defects, macroglossia, neonatal hypoglycaemia and ear abnormalities. There are no strict criteria for a clinical diagnosis of BWS. Generally, it is accepted that the presence of at least three major findings, or two major findings and one minor finding, support the diagnosis¹ (table 2). As in our case, when BWS is suspected, a microdeletion at imprinting centre 1 (IC1) and/or IC2, or a mutation in CDKN1C, confirms the diagnosis. However, if this deletion was present in the absence of the appropriate clinical features a diagnosis of BWS could not be made.

The occurrence of oral polyps in children is probably rare although there are no clear data on their frequency. Polyp itself is a generic term describing a macroscopically visible projection above a mucosal surface. It is thought that the majority of oral polyps in children are due to fibroepithelial hyperplasia, and the aetiology is often unknown, although chronic irritation or inflammation is frequently suspected.¹⁰ Other potential causes of oral polyps in children include skeletal muscle tumours such as rhabdomyomas⁵ or rhabdomyosarcomas,⁶ granular cell tumours,⁷ paragangliomas⁸ and benign lipomatous tumours⁹ such as lipomas, lipoblastomas and hibernomas. It is highly vital for pathologists dealing with oral specimens to be vigilant for

Table 1 Panel of immunohistochemical markers for oral polyps¹¹

Marker	Purpose
Desmin, Muscle specific Actin, HHF35, Myogenin, MyoD1, Myoglobin	Stains muscle origin
S-100 protein	Stains neural origin
Chromogranin A	Stains neuroendocrine origin
Neuron-Specific Enolase (NSE)	Stains muscle or neuroendocrine origin

the important association of clinical and histopathological features, as they might overlook such cases.

This is the first report of an oral polyp occurring in BWS. In the present case, histological analysis revealed a polyp originating from skeletal muscle with hyperplastic characteristics that may have resulted from accelerated growth. The cells showed vacuolation and features that cannot be seen in a rhabdomyoma. Hyperplasia was prominent on the right side where the polyp was found.

Children with BWS are known to have an increased risk of malignancy, particularly tumours of embryonic origin, including rhabdomyosarcomas.² While in this case the histology proved the polyp was benign, in view of the malignant potential in BWS, we recommend any oral polyps in the condition should be excised immediately and sent for histological examination.

In summary, we present a case of an oral polyp being the presenting feature of BWS. GI³ and urethral polyps⁴ have both previously been described in BWS, but there are no previous reports of polyps occurring in the mouth. Clinicians should consider the possibility of BWS in children who present with oral polyps if other signs or symptoms of the syndrome are present. Those who care for children with BWS should also periodically enquire about oral health and ensure any oral polyps are excised.

Figure 2 Immunohistochemical staining to myoglobin antibody (×400).

Table 2 Major and minor findings to support a clinical diagnosis of BWS. It is generally accepted that the presence of at least three major findings, or two major findings and one minor finding, support the diagnosis (adapted from¹)

Major findings	Minor findings
<ul style="list-style-type: none"> ▶ Abdominal wall defects; omphalocele or umbilical hernia ▶ Macroglossia ▶ Macrosomia ▶ Anterior ear lobe creases/posterior helical pits (bilateral or unilateral) ▶ Visceromegaly ▶ Embryonal tumour in childhood ▶ Hemihyperplasia ▶ Cytomegaly of adrenal fetal cortex ▶ Renal abnormalities ▶ Positive family history with BWS ▶ Cleft palate 	<ul style="list-style-type: none"> ▶ Pregnancy-related findings of polyhydramnios, enlarged placenta, thickened umbilical cord ▶ Neonatal hypoglycaemia ▶ Nevus flammeus ▶ Cardiomegaly/structural cardiac anomalies/cardiomyopathy ▶ Characteristic facies ▶ Diastasis recti ▶ Advanced bone age

BWS, Beckwith-Wiedemann syndrome.

Learning points

- ▶ Oral polyps present in children in the early years should be seriously considered as suggestive of a congenital disorder.
- ▶ Clinicians should consider the possibility of Beckwith-Wiedemann syndrome in children who present with oral polyps if other signs or symptoms of the syndrome are present.
- ▶ Effective communication between dental and medical specialists is pivotal to diagnose lesions in the head and neck regions.

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Competing interests None declared.

Patient consent Obtained.

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