

the large number of 'don't knows' with regard to testicle.

We found that those children who used unusual terms for one part of the body would do so for others. These children may actually be using a private family language. An example was the 6 year old who did a 'charlie', 'let polly out', and possessed a 'dilly dat'.

How far the child's language was affected by their parents' region of origin or socioeconomic grouping was not within the scope of the study. We did, however, identify a group of parents who deliberately encouraged the use of the exact anatomical term from the start.

The use of the taboo words was often a source of amusement to both parent and child with the terms

seemingly chosen to enhance this effect (for example 'chuckerella', 'tuppenny', 'tuffies', 'do a soggy' etc). Also, we noted several first names in the children's vocabulary such as 'charlie', 'dick', 'bobs', 'polly', 'fanny', and Auntie Jane, but can only speculate as to their origin. We found little media influence although 'monster munch' (*defecation*) is probably derived from television.

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Mucosal neuroma syndrome—a phenotype for malignancy

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SUMMARY The mucosal neuroma syndrome is characterised by a typical physical appearance, neuromata on tongue and buccal mucosa, and a high risk of developing medullary thyroid carcinoma and phaeochromocytoma. A case is described and the importance of early recognition for prevention of malignancy is stressed.

The mucosal neuroma syndrome, multiple endocrine neoplasia type II^b is a rare, autosomal dominant condition characterised by a typical physical appearance, multiple mucosal neuromas, and a high incidence of medullary thyroid carcinoma and phaeochromocytoma. Patients have coarse facial features, thickened blubbery lips, and a marfanoid habitus. Neuromas can occur on the tongue, buccal mucosa, and eyelids, and throughout the intestine. Disordered bowel function with constipation or diarrhoea is common. Medullated corneal nerves may be visible on slit lamp examination. Mucosal neuromas may be present from infancy and these together with the characteristic physical appearance should alert paediatricians to the diagnosis before malignancy arises. Unfortunately this seldom happens, the diagnosis being made when metastatic

malignant disease has occurred as this case illustrates.

Case history

A 16 year old girl was investigated at a district general hospital for delayed puberty. On examination she had a slender habitus with a high arched palate, and hyperextensible joints. She had coarse facial features with thickened blubbery lips and neuromata on her tongue and buccal mucosa (Figure). Her thyroid was enlarged, with a hard nodule palpable in the right lobe. She was euthyroid. Thickened corneal nerves were visible on slit lamp examination. She was normotensive. Her height was 144.5 cm (less than the 3rd centile), her upper to lower segment ratio was 0.97, and her weight was 26.8 kg (less than the 3rd centile).

The results of endocrine investigations for delayed puberty were normal. She had a chromosome composition of 47, XX with an additional minute centric fragment. This was not considered important as the same abnormality was found in four other healthy family members. Her physical appearance and neuromata suggested, however, multiple endocrine neoplasia type II^b and she was admitted to this hospital for further investigation.

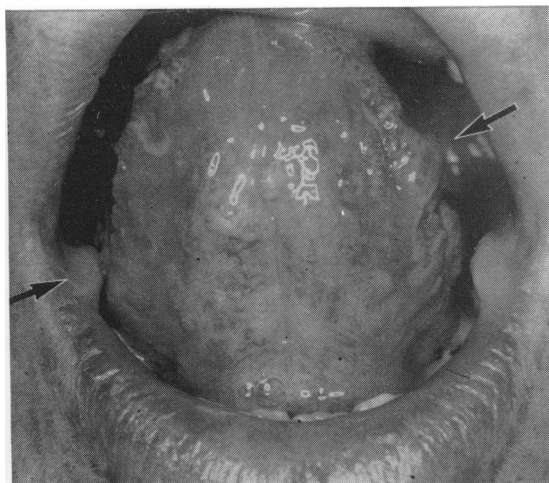


Figure Patient's mouth showing thickened lips and mucosal neuromata (arrowed) on tongue and buccal mucosa.

Her calcitonin value was found to be considerably raised at 131.5 $\mu\text{g/l}$ (normal less than 0.4 $\mu\text{g/l}$). Barium swallow showed the oesophagus to be indented in the region of the thyroid and $\text{Tc}^{99\text{m}}$ thyroid scan showed decreased uptake over the right lobe. Chest radiograph, bone scan, adrenal ultrasound, and barium enema were normal. Full blood count, serum calcium, phosphate, parathormone, gut hormones, and urinary vanilmandelic acid and 4-hydroxy-3-methoxymandelic acid values were also normal.

She underwent thyroid surgery. A highly aggressive tumour was found occupying the right lobe of the thyroid and infiltrating the trachea and lower larynx. Multiple lymph nodes within the carotid sheath and around the thymus were removed and a total thyroidectomy and thymectomy performed. Histology confirmed medullary thyroid carcinoma in the thyroid and all lymph nodes removed. The thymus was normal.

Two months after the operation she is at school but is losing weight. She is receiving 1 α -hydroxycholecalciferol (1 μg) and thyroxine (100 μg) daily. Her calcitonin value remains high at 52.5 $\mu\text{g/l}$.

Her parents (aged 44 and 48 years) and her siblings (aged 22 and 20 years) show no stigmata of multiple endocrine neoplasia type II^b. In this disorder neuromata may rarely be confined to the gut and therefore invisible, but medullary thyroid carcinoma is invariable and leads to early death. In view of this we consider our patient a new mutation.

Discussion

The risk of developing medullary thyroid carcinoma in multiple endocrine neoplasia type II^b approaches 100%.^{1,2} In Khairi's series,² 92.6% had medullary thyroid carcinoma at diagnosis, and 76% had metastases. Medullary thyroid carcinoma is more aggressive and occurs earlier in the type II^b disorder³ than in medullary thyroid carcinoma occurring alone or in multiple endocrine neoplasia type II^a. Therefore total thyroidectomy on the basis of physical appearance alone is recommended,¹ and should certainly be performed if calcitonin values are raised either basally or after provocation tests.⁴

Visible neuromata may be present from a very early age³ and are pathognomonic of this condition. In our patient, neuromata had seemingly been present on the tongue since infancy, but the importance of this finding was not appreciated. Symptoms dated from the neonatal period: she had fed poorly, was floppy, and had severe constipation. At 7 months Werdnig-Hoffmann disease was suspected, but two muscle biopsies were normal. Thereafter her constipation gradually improved and hypotonia resolved by 4 years of age.

Disturbance of bowel function is a very common finding in this disorder.²⁻⁴ Hypotonia is rarer but has been reported.^{3,5} Thus multiple endocrine neoplasia type II^b needs to be considered in the differential diagnosis of Hirschsprung's disease and the floppy baby syndrome.³

We emphasise that the characteristic physical appearance and the presence of neuromata in these patients should be regarded as a marker for occult or potential malignancy.

References

- 1 Norton JA, Froome LC, Farrell RE, Wells SA. Multiple endocrine neoplasia type II^b—the most aggressive form of medullary thyroid carcinoma. *Surg Clin North Am* 1979;**59**: 109-18.
- 2 Khairi MR, Dexter RN, Burzynski NJ, Johnston CC. Mucosal neuroma, pheochromocytoma and medullary thyroid carcinoma: multiple endocrine neoplasia type 3. *Medicine* 1975;**54**: 89-112.
- 3 Moyes CD, Alexander FW. Mucosal neuroma syndrome presenting in a neonate. *Dev Med Child Neurol* 1977;**19**:518-21.
- 4 Graze MD, Spiler IJ, Tachjian AH, et al. Natural history of familial medullary thyroid carcinoma. *N Engl J Med* 1978;**299**:980-5.
- 5 Brown RS, Colle EC, Tashjian AH. The syndrome of multiple mucosal neuromas and medullary thyroid carcinoma in childhood. *J Pediatr* 1975;**86**:77-83.

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