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 deliber-
ately 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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permission to study their patients who seemed to enjoy the
interviews as much as we did.
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Mucosal neuroma syndrome—a phenotype for malignancy

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SUMMARY The mucosal neuroma syndrome is char-
acterised by a typical physical appearance, neuro-
matas 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 IIb is a rare, autosomal dominant
condition characterised by a typical physical appear-
ance, multiple mucosal neuromas, and a high inci-
cence 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. She had coarse
facial features with thickened blubbery lips and
neuromas on her tongue and buccal mucosa (Figure). Her thyroid was enlarged, with a hard
nodule palpable in the right lobe. She was euthy-
roid. 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 summary of endocrine investigations for de-
layed 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 endoc-
rine neoplasia type IIb and she was admitted to this
hospital for further investigation.
Her calcitonin value was found to be considerably raised at 131.5 μg/l (normal less than 0.4 μg/l). Barium swallow showed the oesophagus to be indented in the region of the thyroid and Tc<sup>99m</sup> 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 1α-hydroxycholecalciferol (1 μg) and thyroxine (100 μg) daily. Her calcitonin value remains high at 52.5 μg/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<sup>°</sup>. 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<sup>°</sup> approaches 100%.<sup>1</sup> In Khairi's series,<sup>2</sup> 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<sup>°</sup> disorder<sup>4</sup> than in medullary thyroid carcinoma occurring alone or in multiple endocrine neoplasia type II<sup>°</sup>. Therefore total thyroidectomy on the basis of physical appearance alone is recommended,<sup>1</sup> and should certainly be performed if calcitonin values are raised either basally or after provocation tests.<sup>4</sup>

Visible neuromata may be present from a very early age<sup>3</sup> 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.<sup>2–4</sup> Hypotonia is rarer but has been reported.<sup>3,5</sup> Thus multiple endocrine neoplasia type II<sup>°</sup> needs to be considered in the differential diagnosis of Hirschprung's disease and the floppy baby syndrome.<sup>3</sup>

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**


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