Cretinism and lingual thyroid presenting in an adult

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Summary: A 29 year old woman presented with menorrhagia secondary to hypothyroidism. Subsequent investigation confirmed the presence of a lingual thyroid. Features of cretinism were present despite the late presentation. This unusual case is discussed with reference to previous publications.

Introduction

Lingual thyroid is a rare disorder first described by Hickman in 1869.\(^1\) It has a prevalence of approximately 1 per 100,000 to 300,000 patients\(^2,3\) and 1 in 4000 cases of thyroid disease.\(^4\) It most commonly presents with local symptoms including dysphagia, dysphonia and dyspnœoa.\(^5,6\) Occasionally respiratory difficulty or haemorrhage can occur.\(^7\) At least 15% of patients either present with or are hypothyroid at the time of diagnosis. Female patients predominate in all reported series, ranging from 75 to 89% of the cases.\(^3,5,9\) In this report we present the case of a woman who presented at the age of 29 with menorrhagia and hypothyroidism. Despite the late presentation, she had features to suggest that hypothyroidism had been present from birth. However, local symptoms due to ectopic thyroid tissue were absent.

Case report

A 29 year old Chinese female presented in September 1985 with a 4-year history of menorrhagia. She had emigrated to Hong Kong from Tung Koon county of the Canton Province with her family in 1978. Her growth and development had been abnormal from birth. Although delivery was normal and at full term, developmental milestones were delayed. Her mother noticed that she had a slightly coarser cry than her siblings. She was able to sit unaided only at 7–8 months. At around the same time, she developed a protuberant abdomen. She could speak and walk only at 7 years. Her level of scholastic achievement was poor, she could not perform simple mental arithmetic and could only manage simple household work. Her growth remained stunted, and her menarche was delayed until the age of 20. There was no family history of either thyroid disease or mental retardation. The county from which she emigrated is a coastal area of the South China Sea.

On examination she was short (height 127 cm, well below 3rd centile) with features compatible with cretinism (Figure 1). The upper to lower segment ratio was 69/57. Her head was disproportionately large and limbs short. Body weight was 42.3 kg (10th–25th centile). She was clinically hypothyroid with coarse thick skin, cold peripheries and slow-relaxing ankle and biceps jerks. No thyroid tissue was palpable in the neck nor visible in the buccal cavity. Some secondary sexual development was present (breast development at Tanner Stage 3 and pubic hair at Tanner Stage 4) and there was mild galactorrhoea expressed from the left breast. Pelvic examination done per-rectally showed a normal-sized uterus and no abnormal masses.

Investigation revealed the following findings: serum thyroxine <26 nmol/l (normal 60–140 nmol/l), thyroid stimulating hormone (TSH) 286 mIU/l (normal <4.5), thyroid antibodies negative. Prolactin 1267 pmol/l (normal <877 pmol/l). Haemoglobin (Hb) 7 g/dl with a normochromic normocytic peripheral film. Fasting cholesterol 7 mmol/l (normal 3.6–6.7 mmol/l). Chest X-ray showed marginal cardiomegaly and echocardiography confirmed a moderate pericardial effusion. The pituitary fossa was radiologically normal and epiphyseal fusion was complete. A technetium pertechnetate thyroid scintiscan showed no significant uptake in the neck and a well-defined area of increased uptake at the root of the tongue, indicating the diagnosis of lingual thyroid (Figure 2). The mental age as assessed by the Stanford–Binet Intelligence Scale (SBIS) was 5 years 6 months only.

In view of the severity and apparent long duration of the hypothyroidism together with the low body
weight of the patient, thyroxine treatment was cautiously started using an initial dose of 12.5 µg/day. This has been gradually increased over several months to the current dose of 100 µg/day. Considerable clinical improvement has occurred and the plasma TSH is now normal.

**Discussion**

Approximately 250 cases of lingual thyroid had been reported up to 1977.2 This patient is unusual in that despite the evidence that hypothyroidism had been present since birth or the early postnatal period, the diagnosis was delayed until the complaint of menorrhagia led to its detection at the late age of 29. A protective, closely-knit family environment and inaccessible medical expertise had acted together to prevent earlier recognition and treatment. Technetium pertechnetate scan revealed a lingual thyroid without other co-existing thyroid tissue, as is reported to occur in 70% of cases of lingual thyroid.8,10,11

Adult patients with lingual thyroid may present with hypothyroidism. However, features ofcretinism are then usually absent implying that the ectopic tissue has secreted adequate thyroid hormone during childhood, thus preventing the development of cretinism and leading to a diagnosis of hypothyroidism later in life. Hutchison12 postulated that the age of onset of thyroid insufficiency depended upon the amount of thyroid tissue present.

In children lingual thyroid is an important cause of sporadic hypothyroidism, in one series accounting for 44% of cases in which investigation included an isotope scan.13

The overall prevalence of hypothyroidism in lingual thyroid varies in different series, between 14.5% and 33%.3,4 The exact relationship between ectopic thyroid and hypothyroidism still awaits elucidation. Some have suggested that antibodies directed towards the thyroid cause arrest of descent during early fetal life and poor function.14 Gabr proposed that a metabolic defect in thyroid hormone synthesis might be in some way related to the failure of descent.15 Alternatively, others believe that since the ectopic tissue is frequently hypoplastic, the maldescent may be secondary to the maldevelopment of the gland.16 The issue is reminiscent of the relationship between cryptorchidism and testicular malfunction in which questions still exist as to whether the testis functions poorly because of maldescent or fails to descend completely because it is abnormal.

Treatment of lingual thyroid has been increasingly conservative in recent years. The current trend is to use thyroid hormone in sufficient dosage to suppress TSH stimulation and minimize goitrous enlargement. Sur-
surgery is only indicated if there are complications such as haemorrhage, cystic degeneration, or suspected malignancy. Treatment with radioactive iodine is no longer routinely used and may induce fibrotic change and scarring. In this patient local complications were absent and replacement thyroxine was the only treatment required.

It is noteworthy that the most extensive review of the natural history of lingual thyroid dates from 1936. It is to be hoped that improved medical care universally and the advent of neonatal screening will ensure early diagnosis of associated hypothyroidism. Sadly, this is not yet the case, and with the increased mobility of populations cases like this may continue to appear sporadically in any centre.

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References
