



# Thyroid Dysgenesis

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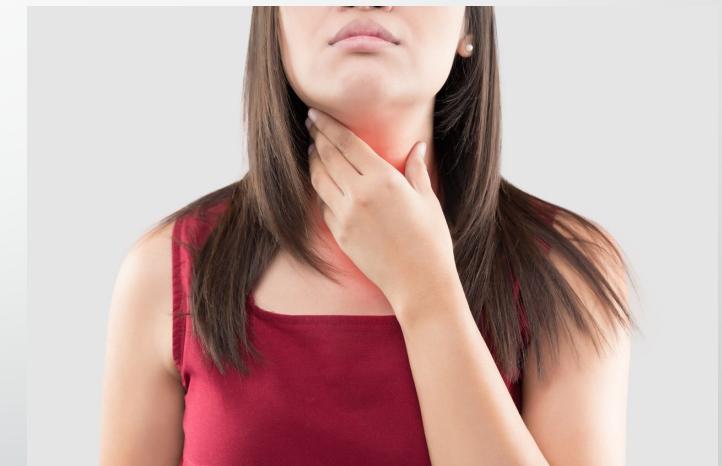
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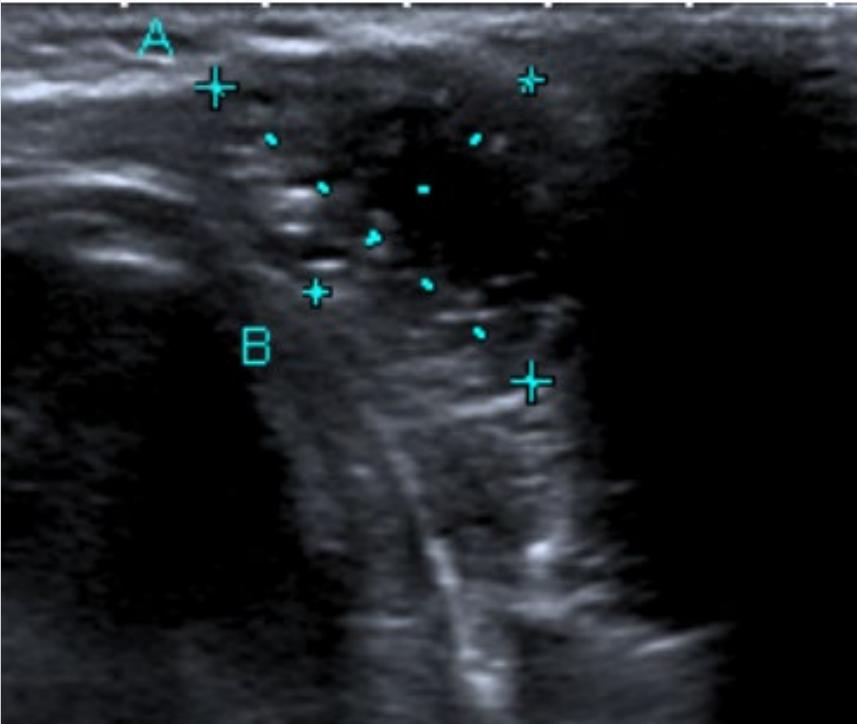
*Grand Rounds*  
October 21<sup>st</sup> 2022

# Case 1

- 28 year old lady
- Presented with a 2 month history of an anterior neck swelling
- Movement on swallow and tongue protrusion
- No family history thyroid disease
- No significant PMH
- TFT normal



# Investigations



USS



CT Neck

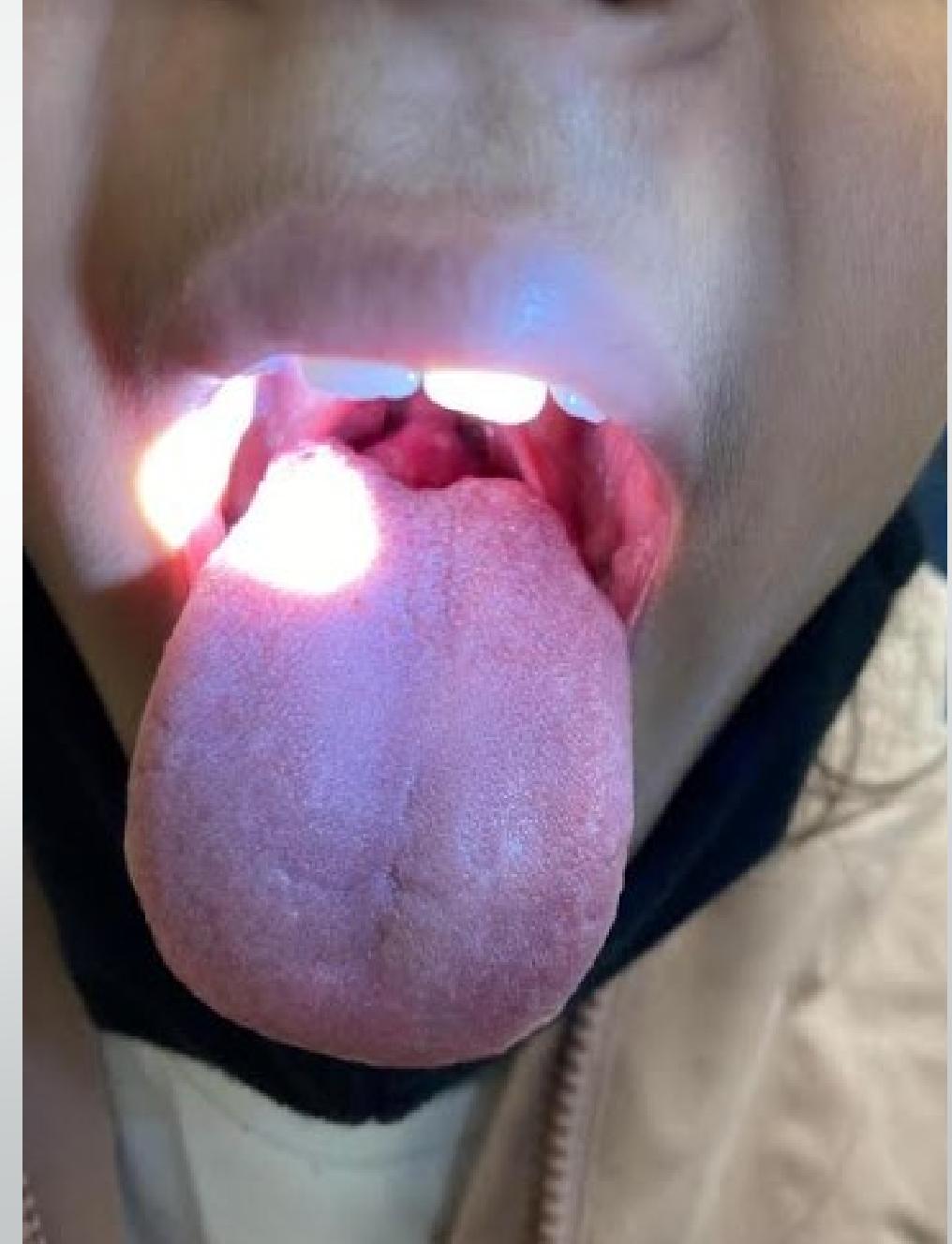
USG FNA: benign cystic fluid

# Management

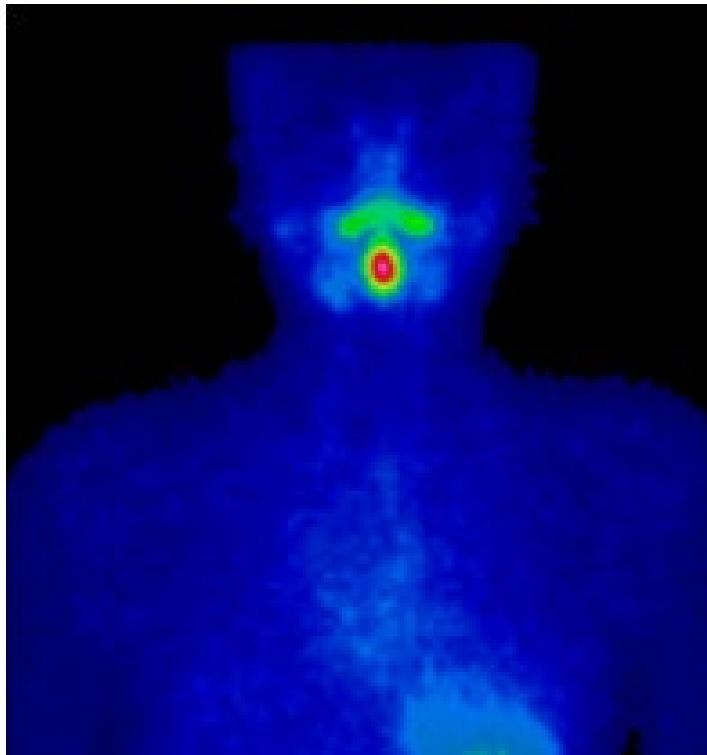
- Proceeded to Sistrunk's procedure
- Post operative histology: cyst containing an 11mm papillary thyroid carcinoma (classical type)
- MDT: no further treatment
- Clinically well, no clinical/radiological recurrence to date

# Case 2

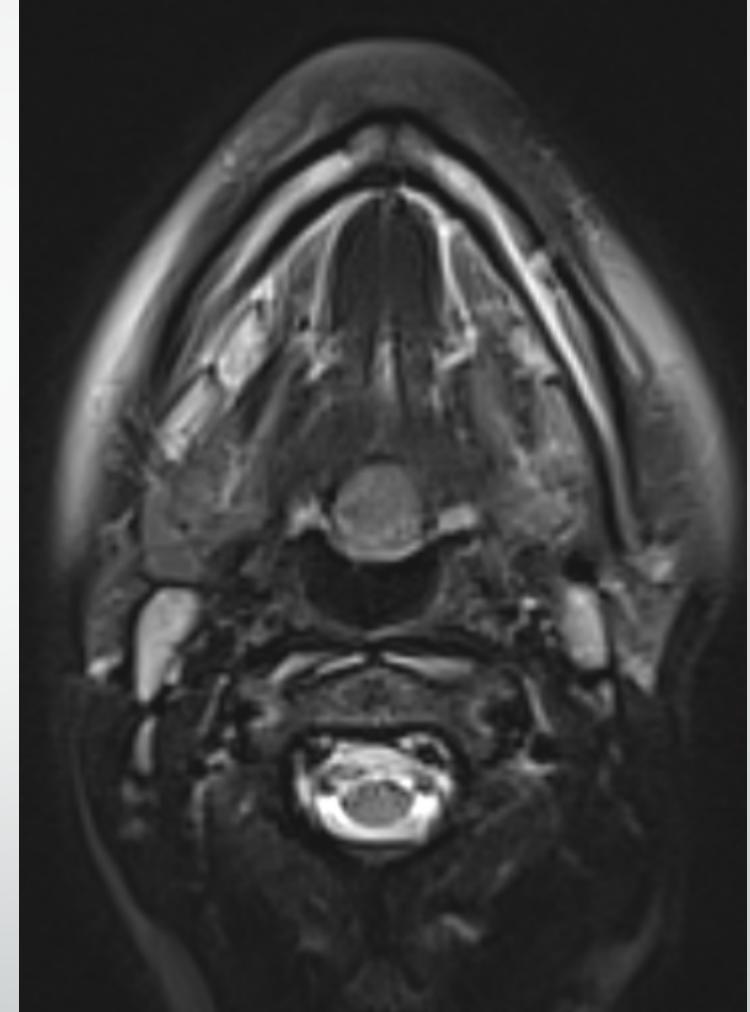
- 11 year old girl
- Posterior tongue swelling
- Unsure of duration
- Otherwise asymptomatic
- No significant medical history
- TSH = 302, T<sub>4</sub> = 3



# Imaging



Technetium uptake scintigraphy



MRI

Lingual thyroid

# Management

- MDT decision: observation
- Surgery not indicated
- Endocrine review for management of hypothyroidism

# Discussion

# Thyroid dysgenesis

- Abnormal thyroid gland development involving various embryonic defects
- Common cause of congenital hypothyroidism
- Many cases are asymptomatic/subclinical
- Gene mutations identified in 5% of cases
  - Linked to defects in gene transcription factors NKX2-1, FOXE1, and PAX-8 which are used in thyroid morphogenesis resulting in abnormal thyroid migration
- 95% of cases are sporadic
- 33% patients hypothyroid

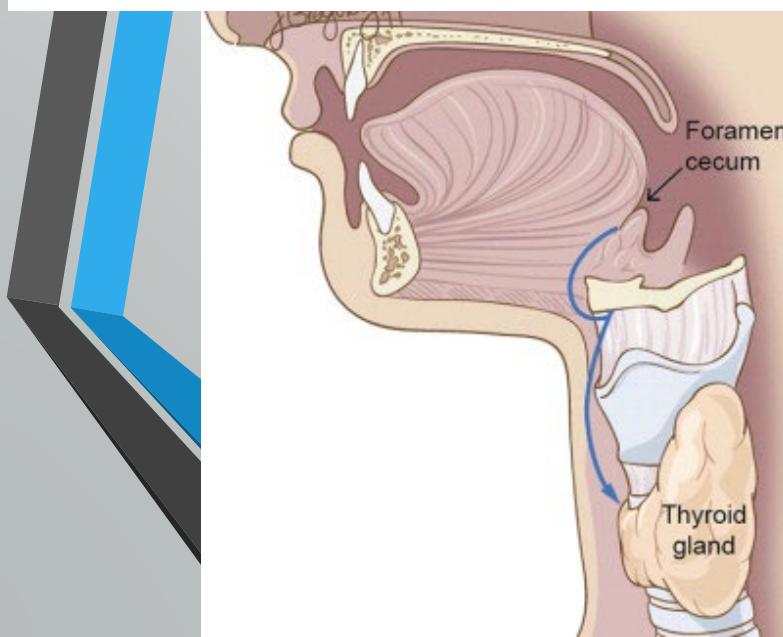
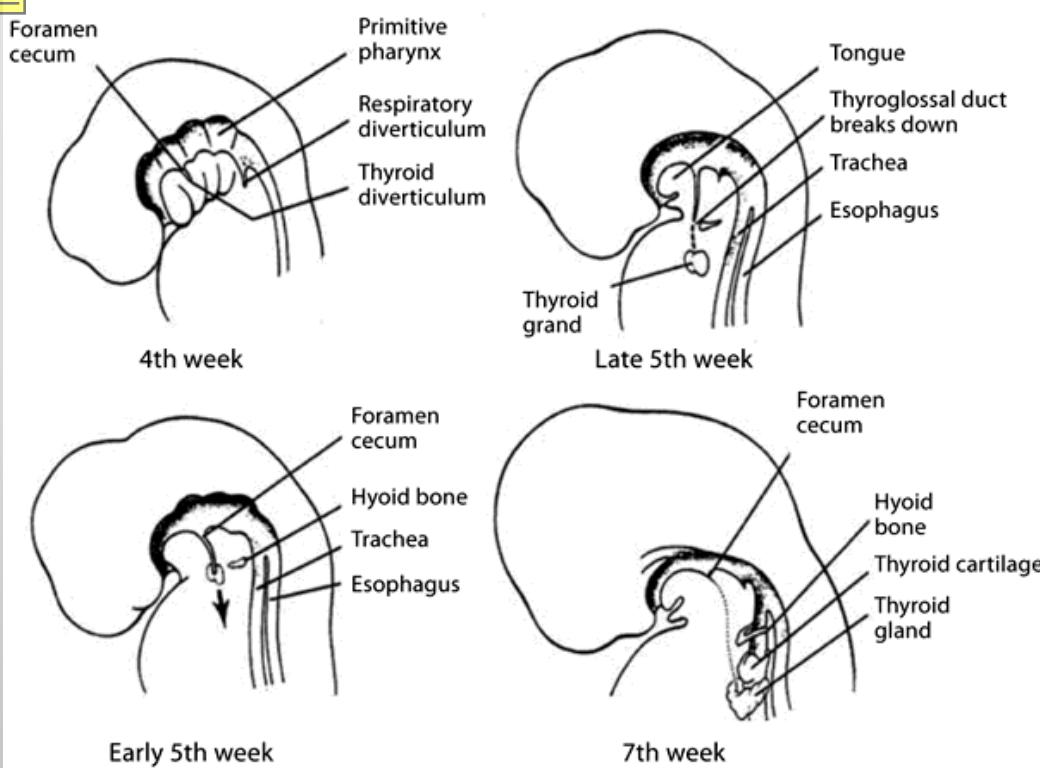


# Manifestations

- Thyroid ectopia
  - Suprahyoid – lingual (most common site), submandibular, skull base
  - Infrathyroid – intratracheal
  - Extracervical – mediastinum, GI tract, adrenal glands
- Dual ectopy
- Thyroglossal duct cyst
- Thyroid hypoplasia/aplasia
- Biochemical status – frequently hypothyroid

# Embryology

- Thyroid is first endocrine gland to develop
- Weeks 3-7 gestation
- Originates from the primitive pharynx and neural crest cells at the foramen caecum between the first and second pharyngeal arches
- Descends from the foramen caecum (at the junction of the posterior and anterior tongue) between 3<sup>rd</sup> – 7<sup>th</sup> week
- Connected to foramen caecum via an endodermal diverticulum (the thyroglossal duct) which typically atrophies prior to full thyroid development
- Passes anterior to the hyoid bone
- Thyroid ectopia is the result of a failure of migration of thyroid, not only along the route of thyroglossal duct but also in subdiaphragmatic organs (e.g. gallbladder, adrenals)



# Thyroid Ectopia

- An ectopic thyroid gland is one which is located in an anatomic location other than the normal position anterior to the laryngeal cartilages
- Prevalence: estimated 1/100,000
- Prevalence in patients with thyroid disease: 1/4000–8000
- 80% of cases in females
- Dual ectopia – ectopic thyroid tissue in 2 locations
- Often asymptomatic but 70% are hypothyroid
- Most commonly occurs along path of thyroglossal duct but can happen anywhere in body
  - Lingual is most common site (at site of foramen caecum)
  - Case reports affecting GI tract, adrenal gland, gallbladder, skull base

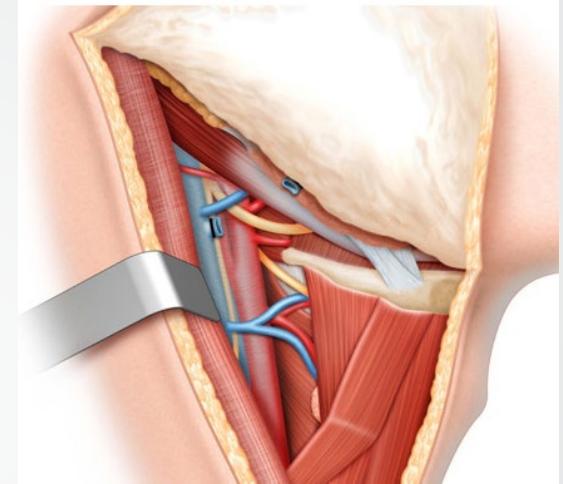
# Lingual thyroid



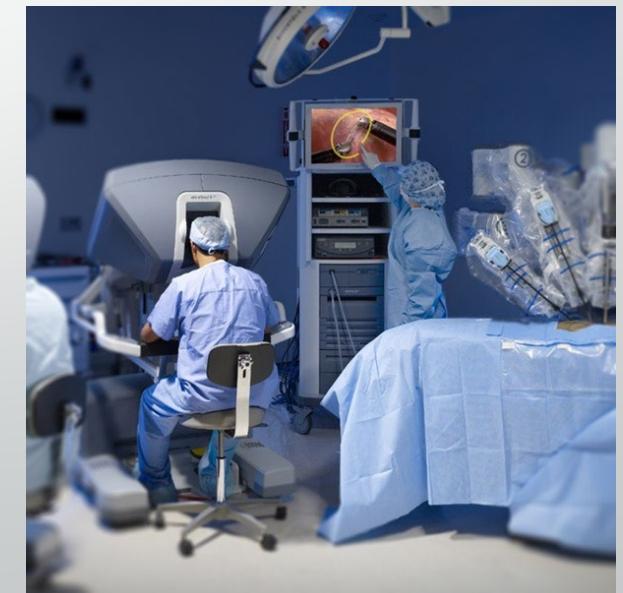
- First described in 1869 (Hickman et al) in a newborn with upper airway obstruction from a lingual thyroid shortly after delivery
- Found at the junction between the anterior 2/3 and posterior 1/3 of tongue
- Most common site of thyroid ectopia (90%)
- However, cadaveric studies have shown microscopic thyroid tissue nests in 10%
- Symptoms: dysphagia, dysphonia, dyspnoea, haemorrhage
- However, most are asymptomatic
- Diagnosis is clinical and radiological
- Technetium 99m scintigraphy most useful scan
- Differentials: dermoid, haemangioma, malignancy, lymphatic malformation, thyroglossal duct cyst
- May be the patients only thyroid tissue
- Cytology can be performed if concern of malignancy

# Management

- If asymptomatic + euthyroid: observation
- Medical management:
  - Correction of hypothyroidism
  - Thyroid suppression therapy
  - Radioiodine ablation
- Surgical:
  - Most patients do not require any surgical intervention
  - Indications: malignancy, uncontrolled hyperthyroidism, significant compressive symptoms
  - Surgical approaches:
    - Lateral pharyngotomy
    - Transhyoid
    - Glossotomy/mandibulotomy
    - Transoral robotic surgery (TORS)

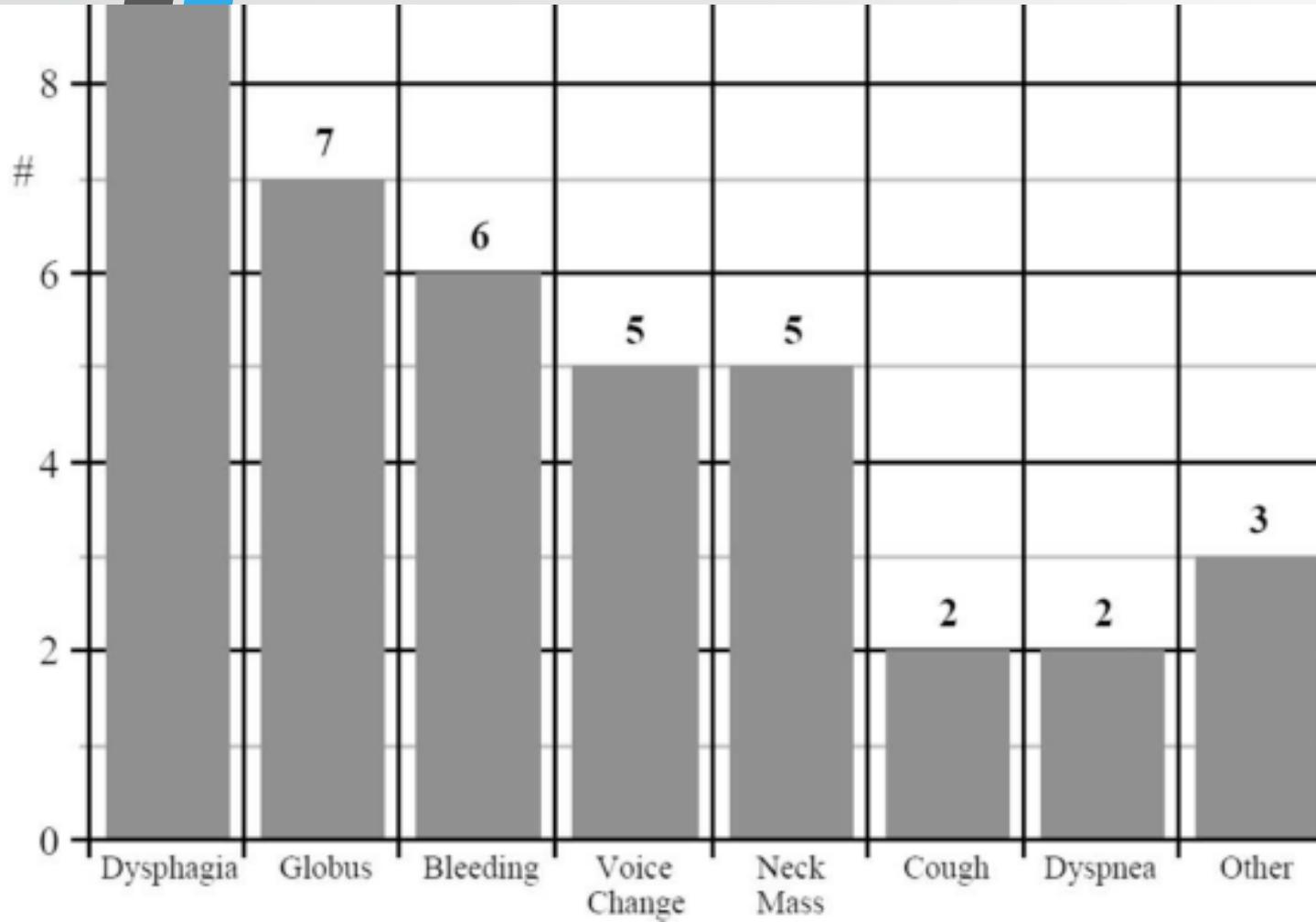


Lateral pharyngotomy

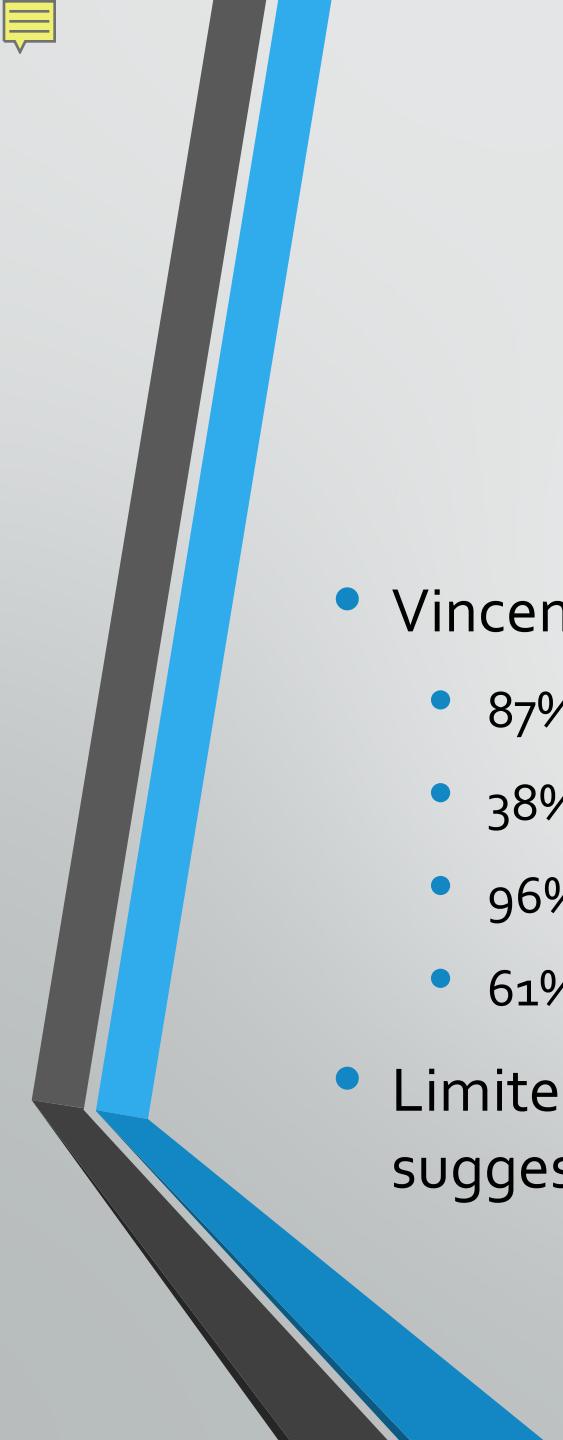


TORS

# Lingual Thyroid Carcinoma



- Very rare – approx. 60 published cases, estimated in ~1% of lingual thyroids
- Differentiating from benign disease:
  - Limited data
  - Clinical suspicion
  - Increasing size
  - Radiology
  - Biopsy



# Lingual Thyroid Carcinoma

- Vincent et al (2019)
  - 87% DTC
  - 38% present with nodal/distant metastases
  - 96% underwent surgical resection
  - 61% adjuvant RAI
- Limited survival data – most case reports with survival outcomes data suggest modest survival after surgery and adjuvant RAI

# Thyroglossal duct cysts (TGDC)

- Cyst formation caused by failure of thyroglossal duct involution during embryological development
- Pathology:
  - Epithelial lining: secretions of cyst contents
  - Can contain salivary gland tissue or thyroid tissue
- Most common congenital neck mass
- Presents as anterior neck swelling
- May present acutely infected from bacterial access via foramen caecum
- Many asymptomatic – cadaveric studies suggest 7% prevalence with at least small cyst/incomplete duct closure
- Location:
  - Suprathyroid: 20-25%
  - At the level of hyoid bone: ~30%
  - Infrathyroid: ~45%

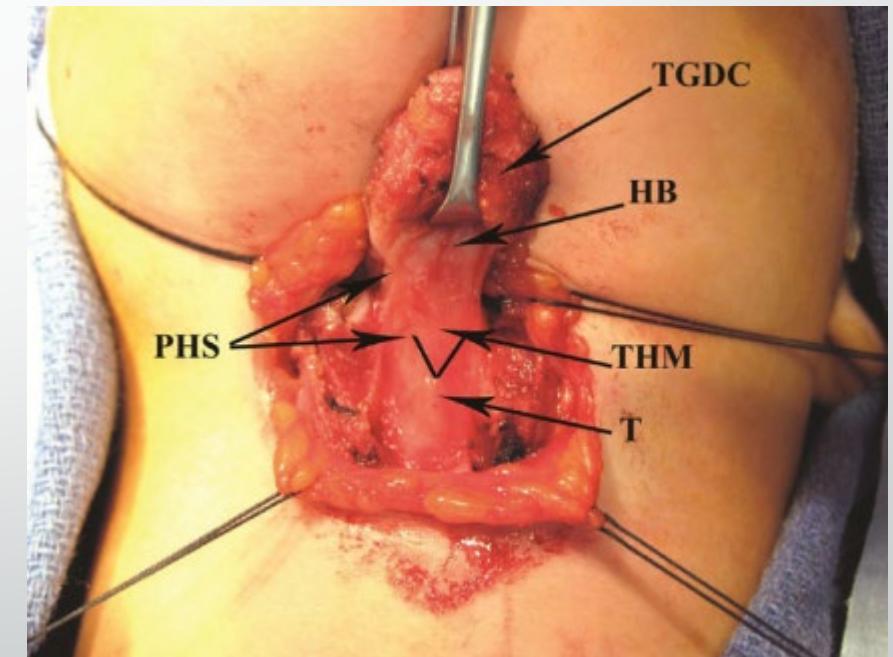


# Workup

- Imaging options:
  - US: useful for differentiating neck masses, presence of thyroid gland
  - Scintigraphy (using Tc-99 m, I-131, or I-123) can detect ectopic thyroid tissue and shows the absence or presence of thyroid in its normal location
  - CT/MRI: can be used for surgical planning if large/atypical location
- Cytology:
  - identification of malignancy: only 53% sensitivity reported
- Biochemistry – TFT's

# Management

- Sistrunk's procedure considered gold standard
- Sistrunk variants including wider dissection of tongue base, central neck dissection etc. not shown to reduce recurrences
- Risk factors for recurrence:
  - Incorrect initial diagnosis (50%)
  - Previous cyst infection (15%)
  - Unusual location (e.g. BOT, lateral neck) (15%)
  - Lack of removal of BOT tissue (2%)
  - Relative inexperience of surgeon (2%)
- May contain patients only source of thyroid tissue





# TGDC carcinoma

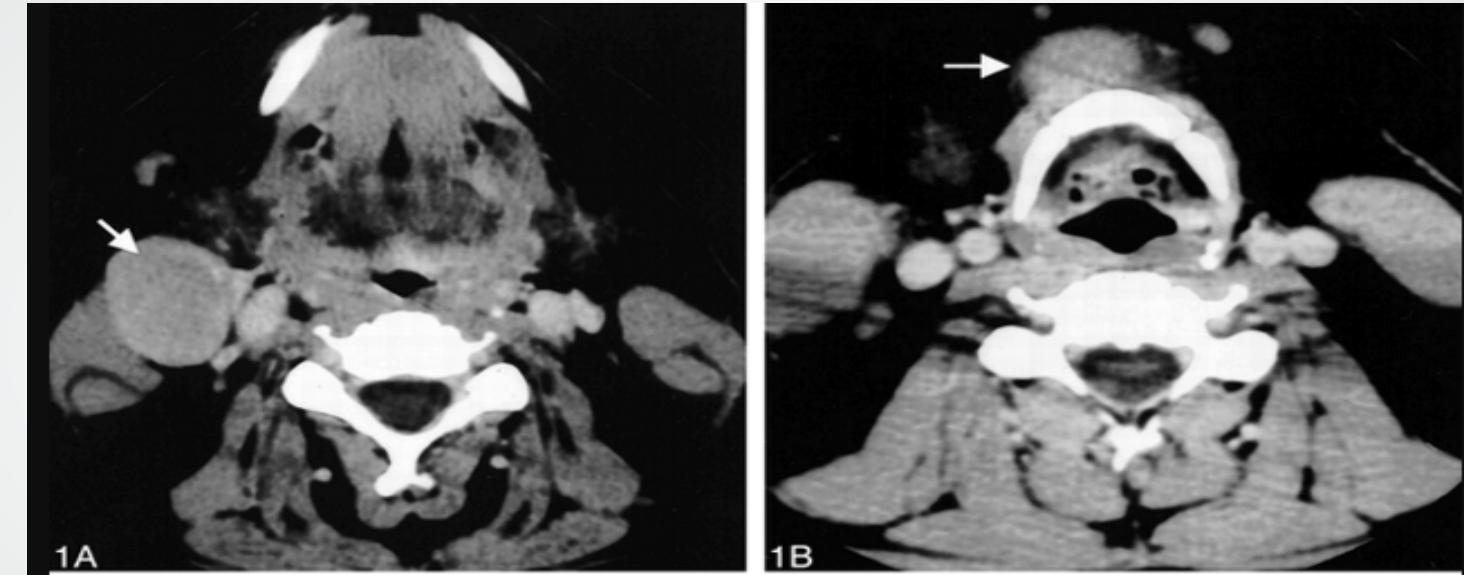
- Occurs in 1% of cysts
- 75% are incidentally diagnosed on histopathology post excision
- Histology:
  - 94% = Thyroid origin (typically PTC)
  - 6% = Squamous
- Differentiation between primary and metastatic deposits can be difficult and is based on clinical and histopathological evaluation
- All require MDT review and US assessment of thyroid gland to assess for synchronous primary (some studies found present in ~60% of cases)

# Preoperative TGDC carcinoma detection?

## Symptoms?

Clinical features, No. %

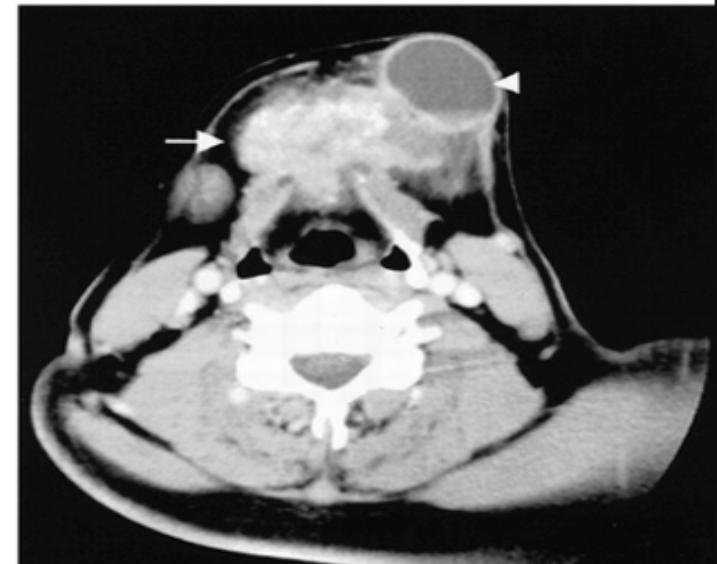
Asymptomatic NM	156 (95.1)
Dysphagia	5 (3.1)
Pain	3 (1.8)



## Tumour size?

Mean tumour size 1.7cm

## Radiological characteristics?



# Management of thyroglossal PTC

- Management based on expert opinion, retrospective reviews, extrapolation from conventional thyroid malignancies
- All require Sistrunk's
- Thyroidectomy?
  - Synchronous thyroid gland tumour, nodal spread
  - Size - >1.5cm
  - Extracystic extension
- Adjuvant RAI
  - Gross extracystic extension
  - Thyroid gland/nodal involvement
  - Presence of aggressive histological variants



# Management of non thyroid thyroglossal malignancies

- Less favourable prognosis overall
- Histopathological assessment essential to differentiate from metastatic disease (e.g. carcinoma of unknown primary)
- SCC
  - 4-6% of cases
  - Arise from epithelium of cyst wall
  - Neck dissection: clinically positive nodes, presence of adverse pathological features (e.g. poorly differentiated)
  - Adjuvant RT: frequently given in case reports. Decision to irradiate generally depends on stage, grade etc.
- Mucoepidermoid
  - Very rare, limited to case reports
  - Arise from ectopic minor salivary gland tissue
  - Should be treated similar to malignant salivary gland tumours of major salivary glands
  - Low threshold for adjuvant RT

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Thank you