Children's Health Ireland

Departments of Respiratory Medicine, Cardiothoracic Surgery & Radiology

Guideline for the Assessment and Management of Asymptomatic Congenital Thoracic Malformations

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Aim

The aim of this guideline is to have a structured, national approach to the assessment and management of asymptomatic congenital thoracic malformations.

Definition of terms

CTM – congenital thoracic malformation which encompasses:
- CPAM – congenital pulmonary airway malformation
- CCAM – congenital cystic adenomatoid / airway malformation
- PS – pulmonary sequestration (extralobar or intralobar)
- CLE – congenital lobar emphysema
- Bronchial atresia
- Bronchogenic cyst

PPB – pleuropulmonary blastoma

Congenital thoracic malformations are best described radiologically in simplified terms in a systematic approach describing the lung parenchyma, bronchial tree, arterial tree and venous tree.

Target Patient Population

This evidence summary applies to asymptomatic infants with an antenatal or postnatal diagnosis of a congenital thoracic malformation. It does not deal with the management of symptomatic infants and does not address antenatal care.

Target Users

This guide is directed at health-care professionals engaged in the care of neonates and children with a diagnosis of a congenital thoracic malformation. This includes obstetricians, neonatologists, respiratory paediatricians, paediatric radiologists and paediatric thoracic surgeons.

Background

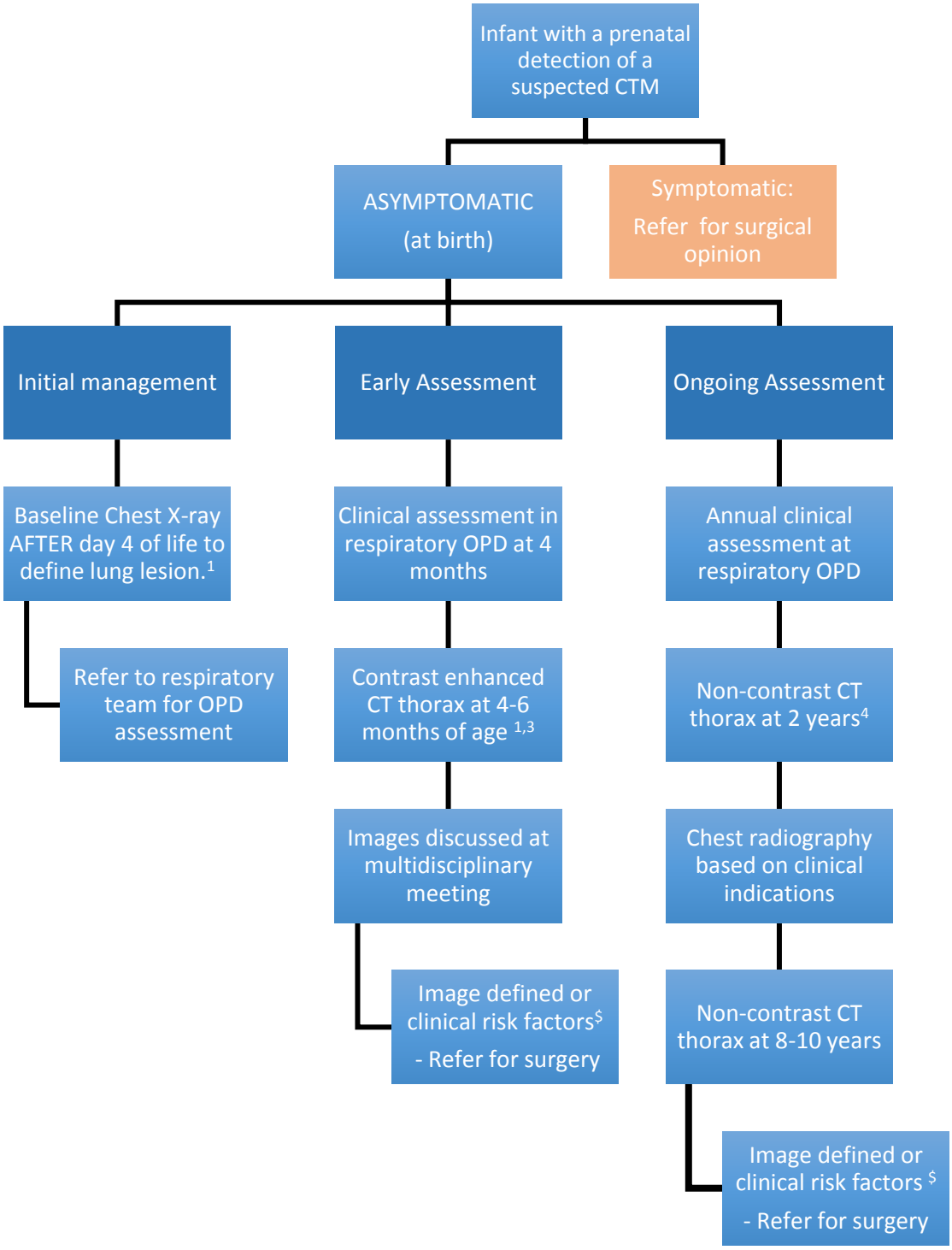
The management of congenital thoracic malformations is controversial varying from resection of all lesions irrespective of symptoms in some units to conservative management of asymptomatic lesions in other units. The evidence base in the literature to guide decision-making is level IV providing grade 3-4 recommendations. This guideline is based on the evidence collated from a systematic review by the American Paediatric Surgical Association in June 2017 and relevant articles published since this review.

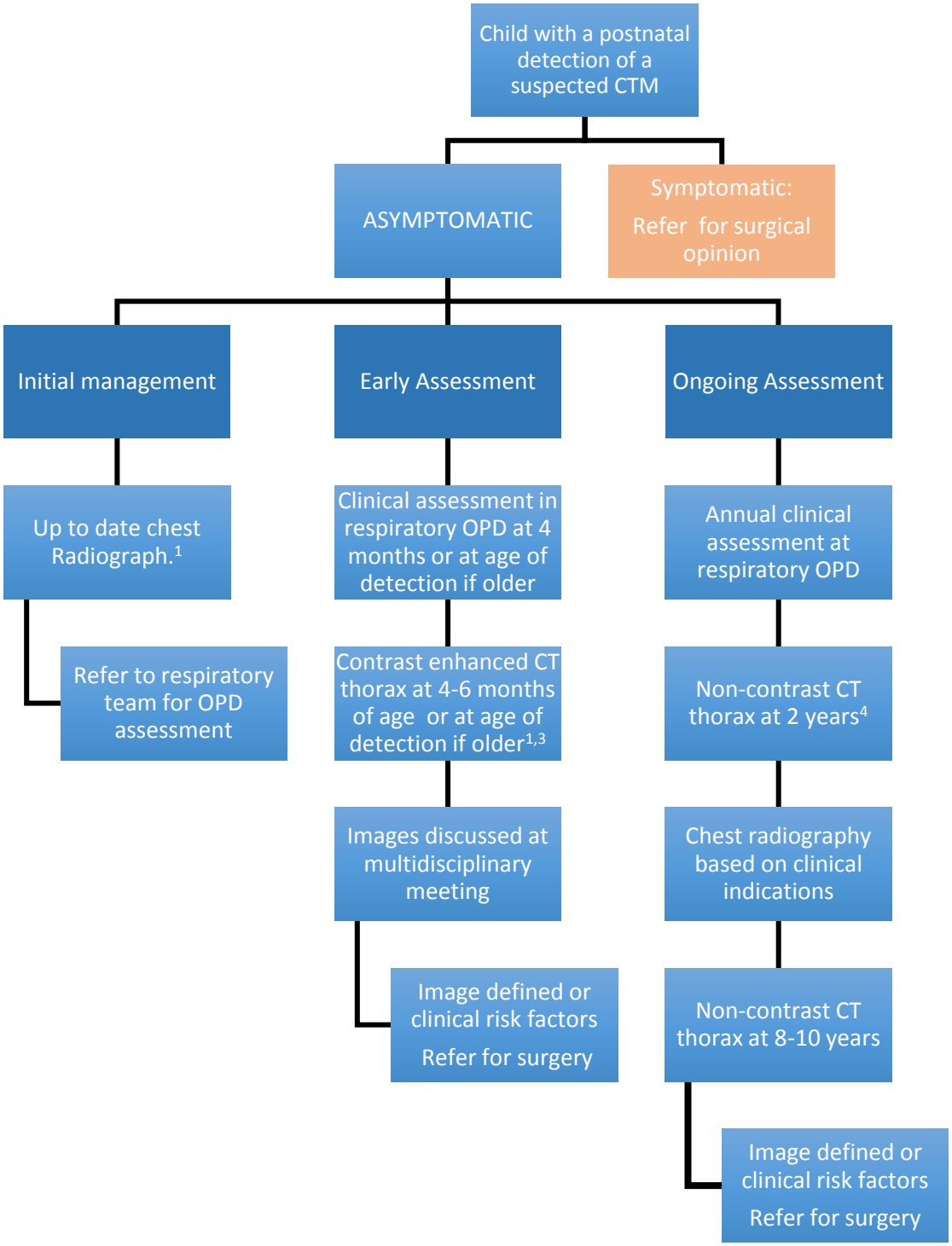
Assessment

This guideline is for asymptomatic infants only. If at any stage there are clinical concerns please discuss with the respiratory paediatrician on call.

Potential problems with congenital thoracic malformations
- Recurrent respiratory tract infection
- Mass effect
- Pneumothorax
- Some can be indistinguishable from a Pleuropulmonary Blastoma (a variably aggressive childhood malignancy)
- arteriovenous shunting

Management


Infant with a prenatal detection of a suspected CTM
ASYMPTOMATIC (at birth)
Symptomatic: Refer for surgical opinion
Initial management
Early Assessment
Ongoing Assessment
Baseline Chest X-ray AFTER day 4 of life to define lung lesion.[1]
Refer to respiratory team for OPD assessment
Clinical assessment in respiratory OPD at 4 months
Contrast enhanced CT thorax at 4-6 months of age [1,3]
Images discussed at multidisciplinary meeting
Image defined or clinical risk factors[$] - Refer for surgery
Annual clinical assessment at respiratory OPD
Non-contrast CT thorax at 2 years[4]
Chest radiography based on clinical indications
Non-contrast CT thorax at 8-10 years
Image defined or clinical risk factors [$] - Refer for surgery


Child with a postnatal detection of a suspected CTM
ASYMPTOMATIC
Symptomatic:
Refer for surgical opinion
Initial management
Early Assessment
Ongoing Assessment
Up to date chest Radiograph.[1]
Refer to respiratory team for OPD assessment
Clinical assessment in respiratory OPD at 4 months or at age of detection if older
Contrast enhanced CT thorax at 4-6 months of age or at age of detection if older[1,3]
Images discussed at multidisciplinary meeting
Image defined or clinical risk factors
Refer for surgery
Annual clinical assessment at respiratory OPD
Non-contrast CT thorax at 2 years[4]
Chest radiography based on clinical indications
Non-contrast CT thorax at 8-10 years
Image defined or clinical risk factors
Refer for surgery

Imaging or Clinical Risk Factors

Imaging Features that may trigger potential surgical intervention[2]	• Multilobar or bilateral involvement • complex cyst - septated or containing solid elements • Mediastinal shift • CTM occupying an entire lobe • Pneumothorax and/or pleural effusion • not seen on antenatal scan
Clinical features that may trigger surgical intervention[1,2,3,4]	• Infection • Pneumothorax and/or pleural effusion • Parental concern • Family history or known genetic markers of PPB
Features that may trigger intervention at Follow-up CT[1]	• Change to structure • New lesions
Imaging features on CT suggestive of Benign CTM rather than PPB[2]	• Any hyperinflated region • Visible feeding vessel

Imaging findings that do not require follow up from a neoplastic perspective:
Isolated congenital lobar emphysema, sequestration, bronchial atresia/focal hyperinflation with no cystic change

Management Issues where evidence is lacking and reasons for guideline decisions

- Chest radiograph at ≥ 4 days → radiographs performed in the first few days postnatally are often non-diagnostic due to persistent fluid filling the CTM therefore to best define the lung lesion a CXR after day 4 of life is more informative
- Initial CT scan at 4-6 months → will allow time for CTM to clear any residual fluid and increased lung maturity and patient bodyweight will enable a more diagnostic scan. This should be an arterial phase contrast CT under GA with inspiratory/expiratory cuts.
- Non-contrast CT at 2 years → Cook et al performed their 2nd scan at 5 years however as the majority of PPBs are diagnosed in the first 2-3 years of life, a 2 year CT scan is felt to be more likely to detect an early PPB[4].
- Non-contrast CT scan at 8-10 years → give a final morphological appearance of the CTM. Case discussed at MDT. Clinical risk factors identified, patient referred for surgery. No clinical risk factors identified, patient discharged.
- Chest radiographs → as required on a clinical basis. No evidence exists for routine chest radiograph imaging.

Role of DICER1 mutation

- Dicer mutation is a known risk factor for the development of PPB among other tumours.
    - Ovarian sex cord tumours / testicular tumours
    - Renal
        - Cystic nephroma
        - Renal sarcoma
        - Wilms tumour
    - Brain
        - Pineoblastoma
        - Pituitary blastoma
    - Nodular hyperplasia of the thyroid and thyroid cancer
    - Nasal chondromesenchymal hamartomas
    - Ciliary body medulloepithelioma
    - Genitourinary (bladder, cervix) embryonal rhabdomyosarcoma
    - Intestinal polyps

- No evidence-based guideline exists regarding routine screening for DICER1 mutations in those presenting with a CTM alone.
- A recent publication by Schulz et al recommends screening for DICER1 in children with a CTM however this recommendation is based on "a consensus of expert opinion and current literature"[4].
- DICER1 mutational analysis should be considered in those with a CTM and a family history of lung cysts, PPB or other tumour consistent with DICER1 mutation. Consideration should also be made for the consequences of genetic testing. The decision to proceed with DICER1 mutation analysis should be made at the MDT in conjunction with clinical genetics. Refer to clinical genetics for cascade screening.

Companion Documents

References

Literature Search Strategy