MICROCEPHALY

- Present at birth and occurs in approximately 3 per 10,000 births
- also known as primary microcephaly
- Defined as head circumference [occipital frontal circumference (OFC)] <2nd centile on the growth chart (severe if <0.4th centile)
- Check whether microcephaly is symmetrical (proportionate to body weight) or asymmetrical
- May indicate significant underlying structural brain abnormality, genetic or metabolic condition, or congenital infection
- Does not necessarily predict functional outcome but can be associated with seizures, cerebral palsy, developmental delay, hyperactivity depending on underlying aetiology

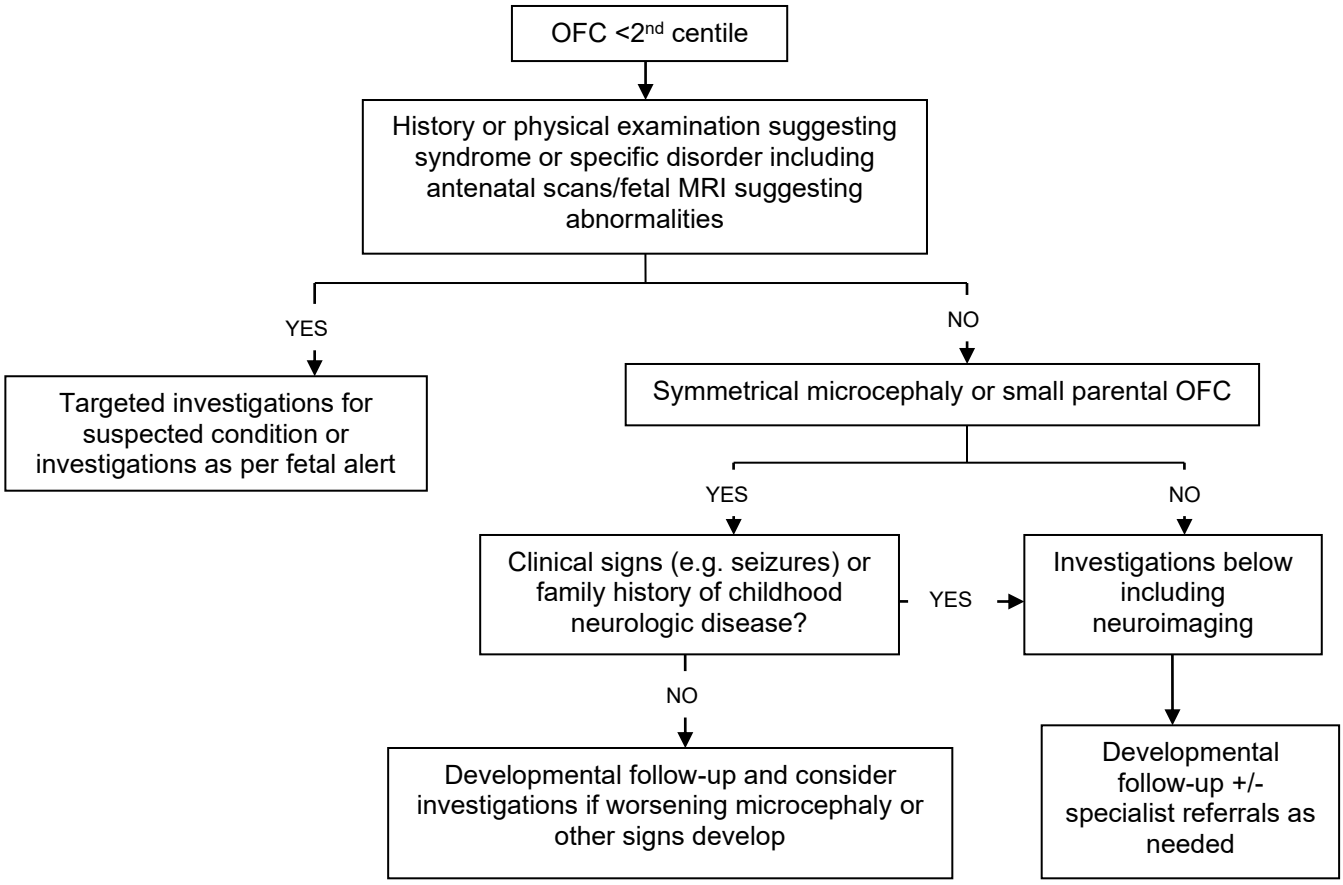
CAUSES

- Aetiology unknown in 40% of cases
- Known aetiology can be broadly categorised as due to premature fusion of sutures (i.e. craniosynostosis) or poor brain growth
- Causes include:
- genetic or chromosomal anomalies e.g. Smith-Lemli-Opitz syndrome, trisomy 13, trisomy 18, trisomy 21, Cornelia de Lange syndrome, Rett syndrome, Cri du chat syndrome, Williams syndrome
- structural or neuroanatomical abnormalities in the brain
- metabolic e.g. pyruvate dehydrogenase deficiency, serine biosynthesis disorder
- intrauterine infections e.g. SCORTCH (syphilis, cytomegalovirus, 'other', rubella, toxoplasmosis, chickenpox, herpes simplex virus and blood-borne viruses), Zika virus, varicella zoster virus, human immunodeficiency virus, lymphocytic choriomeningitis virus (LCMV)
- teratogens e.g. alcohol, cocaine, anti-epileptic drugs, lead/mercury intoxication, radiation
- stroke or death of a twin in-utero
- maternal diseases e.g. hyperphenylalaninaemia, anorexia nervosa, placental insufficiency, hypothyroidism, malnutrition in pregnancy

MANAGEMENT

- Detailed history including:
- prenatal
  - maternal medical history
  - medication/substance misuse exposure in pregnancy
  - antenatal blood and ultrasound scan results
  - travel history of both parents in last 6 months before/during pregnancy
  - family history of inherited conditions
- perinatal
  - gestational age
  - birth weight
  - need for resuscitation at birth
  - risks for infection
- Physical examination
- assess for dysmorphic features
- measure and plot head circumference and weight on growth chart
- where feasible, measure OFC 24 hr after birth to account for effects of moulding, caput succedaneum and cephalhaematoma

INVESTIGATIONS



Viral screen: SCORTCH screen (including urine CMV)

- If mother had negative SCORTCH screen in pregnancy to investigate microcephaly, this should suffice
- if mother travelled to endemic countries in pregnancy or ≤6 weeks before pregnancy (6 months for male partners), consider Zika virus screen

Microarray

- 44% of congenital microcephaly associated with genetic aetiology

MRI head

- Detects abnormalities in approximately 80% of severe microcephaly
- Cranial USS has limited role as excludes mainly gross cortical malformation

Metabolic tests

- Not routinely indicated as first line – metabolic disorders are more likely to cause secondary microcephaly
- Ensure Guthrie test done and results are normal (checks for some inborn errors of metabolism)
- If suspected, toxin screen for heavy metals e.g. lead, arsenic and mercury

TREATMENT

- Management depends on severity and underlying cause
- Severe cases more likely associated with imaging abnormalities or development delay and support services e.g. occupational or physical therapy, speech and language can optimise outcomes
- If associated with seizures (see **Seizure** guideline), anticonvulsants may be required