Microcephaly

Definition / Supporting Information

Microcephaly refers to a head size ≥ 3 standard deviations below the mean, below 0.4th centile.

Micrencephaly refers to having a small brain leading to a small head size.

Craniosynostosis is premature closure of sutures (with normal brain growth).

- Measure head circumference with a flexible, non-stretchable tape
  - Measure above the supraorbital ridges and across the occipital prominence
  - Repeat so three measurements are taken
  - Plot measurements on standardised charts (see UK-WHO growth charts (pdf) for boys and girls)
    - Adjust head circumference for prematurity until 2 years of age (in infants with birth weight < 1000 g, use corrected age until 3 years of age, or until growth has caught up to normal)
    - Use charts specific to special populations when indicated (eg, Down's syndrome, Williams' syndrome, achondroplasia, very-low-birth-weight infants)
    - Include head circumference at birth and any previous measurements available in Red Book to review velocity of head growth, and whether acquired or congenital
- Measure head circumferences of other family members if possible

Keywords / also known as: reduced head size, small head circumference

Essential History

Ask about:

- Prenatal history
  - Explore potential toxin exposure (alcohol, drugs)
  - Review maternal health during pregnancy
  - Review records and assess the likelihood of intrauterine infections
  - Review records and assess the likelihood of placental insufficiency
  - Discuss psychosocial factors, including history of prenatal care and maternal education
- Medical history, including:
  - Previous infections, such as meningitis

- Review of systems, including:
  - Full dietary history
- Family history
  - Construct a pedigree, noting genetic syndromes, miscarriages, and microcephaly
- Developmental history
  - Include a chronology of developmental milestones
  - Assess the child's current function and behaviour

'Red Flag' Symptoms and Signs

Ask about:

- Symptoms of raised intracranial pressure
  - Vomiting
  - Lethargy
  - Headaches (if old enough to enunciate)
- Plateauing, loss (regression), or delay of developmental skills
- Seizures

Look for:

- Signs of raised intracranial pressure
  - Hypertension
  - Bradycardia
  - Irregular respirations
- Head circumference crossing centiles
- Abnormal head shape, bony ridges or premature fusion of the fontanelles
  - Craniosynostosis
- Evidence of congenital malformations
- Evidence of involvement of other organ systems, such as:
  - Ocular abnormalities (eg, colobomas, cataracts)
  - Congenital heart disease
  - Genitourinary abnormalities (eg, cryptorchidism)
- Abnormalities on full neurological examination
  - Cranial nerve dysfunction
  - Papilloedema
  - Asymmetries or abnormalities in muscle tone, posture, strength, and reflexes

Differential Diagnosis / Conditions

- Asymptomatic familial microcephaly
  - Small head size in context of normal development, normal neurological examination, and family history

- Genetic causes (primary microcephaly)
  - Small brain / abnormal brain development
    - Microcephaly vera ("true microcephaly")
      - Brain size usually 3 standard deviations below the mean
      - Brain architecture grossly normal
      - Patients almost always have learning disabilities but have otherwise unremarkable neurological examination
      - May see sloping forehead and prominent ears
    - Microcephaly with severe neurological impairment (seizures, spasticity, and / or global developmental delays)
      - More common than microcephaly vera
      - Neuroimaging may identify abnormal brain architecture.
  - Genetic syndromes
- Environmental causes (secondary microcephaly)
  - Intrauterine environment / perinatal timeframe
    - Teratogens
    - Foetal alcohol syndrome
    - Untreated maternal phenylketonuria
    - Intrauterine infections (eg, TORCH (toxoplasmosis, other agents, rubella, cytomegalovirus, and herpes simplex))
    - Intrauterine irradiation
    - Hypoxic–ischaemic encephalopathy
  - Infections such as meningitis
  - Severe malnutrition
  - Traumatic brain injury, including shaken baby syndrome
- Craniosynostosis
  - Multiple sutures
  - Problem of skull, not brain

Investigations

To be undertaken by specialist practitioners (eg, Paediatric / Paediatric Neurology / Neurosurgery Team(s)):

- Laboratory evaluation as guided by clinical suspicion
- In children with possible intrauterine infection
  - Test for perinatal infections
    - Toxoplasmosis
    - Rubella
    - Cytomegalovirus
    - Syphilis
    - HIV

        - Herpes
    - Ophthalmic and audiological evaluations
- If a genetic syndrome is suspected, perform a focused evaluation, consulting experts in genetics as needed
- Magnetic resonance imaging (MRI)
    - Grey and white matter disease
    - Migration defects, such as lissencephaly, pachygyria, and polymicrogyria
- Computed tomography
    - Intracranial calcifications due to intrauterine infections
    - Skull abnormalities, as seen with premature fusion of cranial sutures
    - Ventricular system abnormalities
- Radiography of the skull
    - Can show intracranial calcifications
    - Can show characteristic bone findings in craniosynostosis
- Ultrasonography
    - Useful in neonates / young infants with open fontanelles

When to Refer

Refer (arrange emergency transport) to specialist practitioners (eg, Emergency Department / Paediatric / Paediatric Neurology / Neurosurgery Team(s)) if:

- Signs of increased intracranial pressure
- Changes in mental status (see Altered Conscious Level)
- Any other acute neurological deterioration or abnormal neurological examination
- Seizures

Refer to specialist practitioners (eg, Paediatric / Paediatric Neurology / Neurosurgery Team(s)) if:

- Head circumference > 3 standard deviations below the mean
- Deceleration of head growth with head circumference crossing centiles
- Dysmorphic features
- Regression in motor, language, and social skills
- Suspected craniosynostosis

‘Safety Netting’ Advice

- Advise parents to seek medical advice if any ‘red flag’ symptoms develop.

Patient / Carer Information

Please note: whilst these resources have been developed to a high standard they may not be specific to children.

- Craniosynostosis (Web page), the NHS website

Resources

Suggested Resources

Please note: these resources include links to external websites. These resources may not have national accreditation and therefore PCO UK cannot guarantee the accuracy of the content.

UK-WHO growth charts (pdf) for boys and girls.

Microcephaly (Web page), Contact a Family.

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National Organization for Rare Disorders (Web site), NORD.

OMIM—Online Mendelian Inheritance in Man (Web site), McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University (Baltimore, MD), and National Center for Biotechnology Information, National Library of Medicine (Bethesda, MD).

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