



Fifteen-minute consultation on the infant with a large head

Arnab Seal

Correspondence to

Dr Arnab Seal, Leeds Community Healthcare NHS Trust and Leeds Teaching Hospitals NHS Trust, Children's Services, Wortley Beck Health Centre, Ring Road, Lower Wortley, Leeds, West Yorkshire LS12 5SG, UK; arnab.seal@nhs.net

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ABSTRACT

An infant with a large head (2.5 SDs above normal for weight and gender or above 99.6th centile for age) is a common clinical presentation. Usually, it is due to benign isolated macrocephaly or familial macrocephaly (FM) where some close family members are similarly affected¹; neither condition requires any further intervention. However, there are a few important underlying causes the clinician needs to actively consider and investigate when indicated before reassuring parents. These considerations include whether there is any associated developmental disorder or suggestion of a syndromic association or evidence of raised intracranial pressure (ICP).

CLINICAL APPROACH

In most instances, a careful history and examination either clarifies the diagnosis; for example, an ex-preterm child developing hydrocephalus. In other instances, it provides pointers for further investigation and management, for example, arranging genetic tests in a child presenting with macrocephaly and multiple *café au lait spots* suggesting the possible diagnosis of neurofibromatosis (NF) Type 1.

The following considerations are helpful.

1. Is the head actually large? The clinician should accurately measure and plot occipito-frontal circumference (OFC) using appropriate non-stretch tape measure. The OFC extends from the most prominent part of the glabella to the most prominent posterior area of the occiput or the largest measureable circumference. The OFC can be affected by thick hair and cranial bone deformations or hyperostosis. Height and weight should be plotted simultaneously as the OFC may be within normal centiles, but significantly discordant with body growth. If OFC is within normal centiles, but the head seems apparently large, then consider late intrauterine

Box 1 Conditions associated with higher risk of presenting with a large head in infancy

- ▶ History of other immediate family member/s having asymptomatic large head
- ▶ Ex-preterm baby with history of intra-ventricular haemorrhage
- ▶ Previous history of subdural haemorrhage
- ▶ Neonatal meningitis
- ▶ Presence of non-cranial congenital anomalies can increase risk of brain anomalies and/or hydrocephalus, for example, spina bifida
- ▶ Family history of genetic syndromes associated with macrocephaly for example, neurofibromatosis type 1
- ▶ Relevant family history of developmental problems, for example, autism or learning disability

growth retardation, skeletal dysplasias for example, osteogenesis imperfecta, and conditions causing frontal bossing, for example, rickets.

2. Are there any serial measurements? Often the birth OFC and subsequent measurements are available from neonatal records and parent-held child health surveillance records. Is the OFC crossing centiles upwards? This does not necessarily

Table 1 Signs and symptoms of raised intracranial pressure (ICP) in infancy

Symptoms of raised ICP	Signs of raised ICP
Irritability	Bulging fontanelle
High-pitched cry	Splayed sutures
Poor feeding	OFC crossing centiles
Vomiting	Sunsetting eyes
Decreased alertness	Prominent scalp veins
Developmental delay	Recent onset squint
Seizures	Lower limb spasticity
	Hyperreflexia
	Papilloedema

OFC, occipito-frontal circumference.

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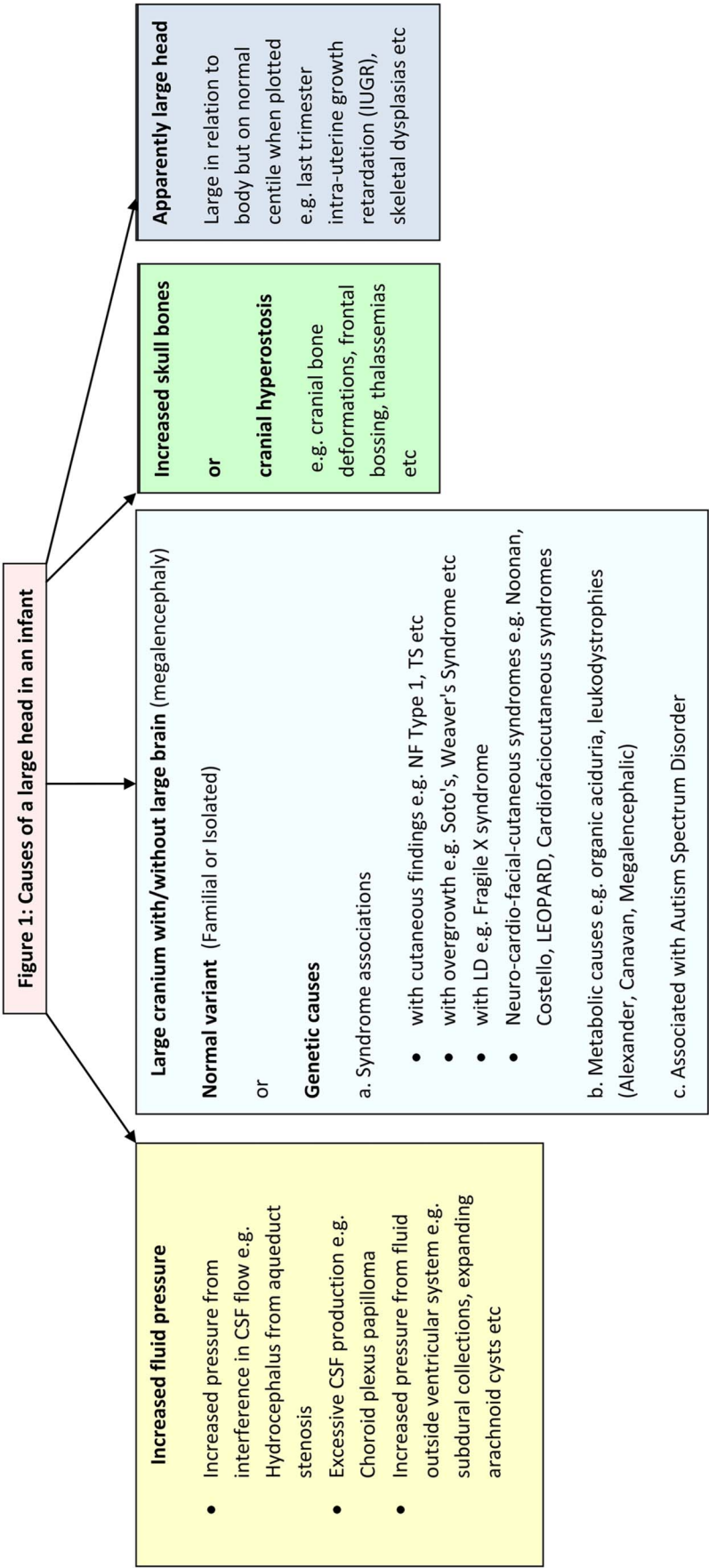


Figure 1 Causes of a large head in an infant.

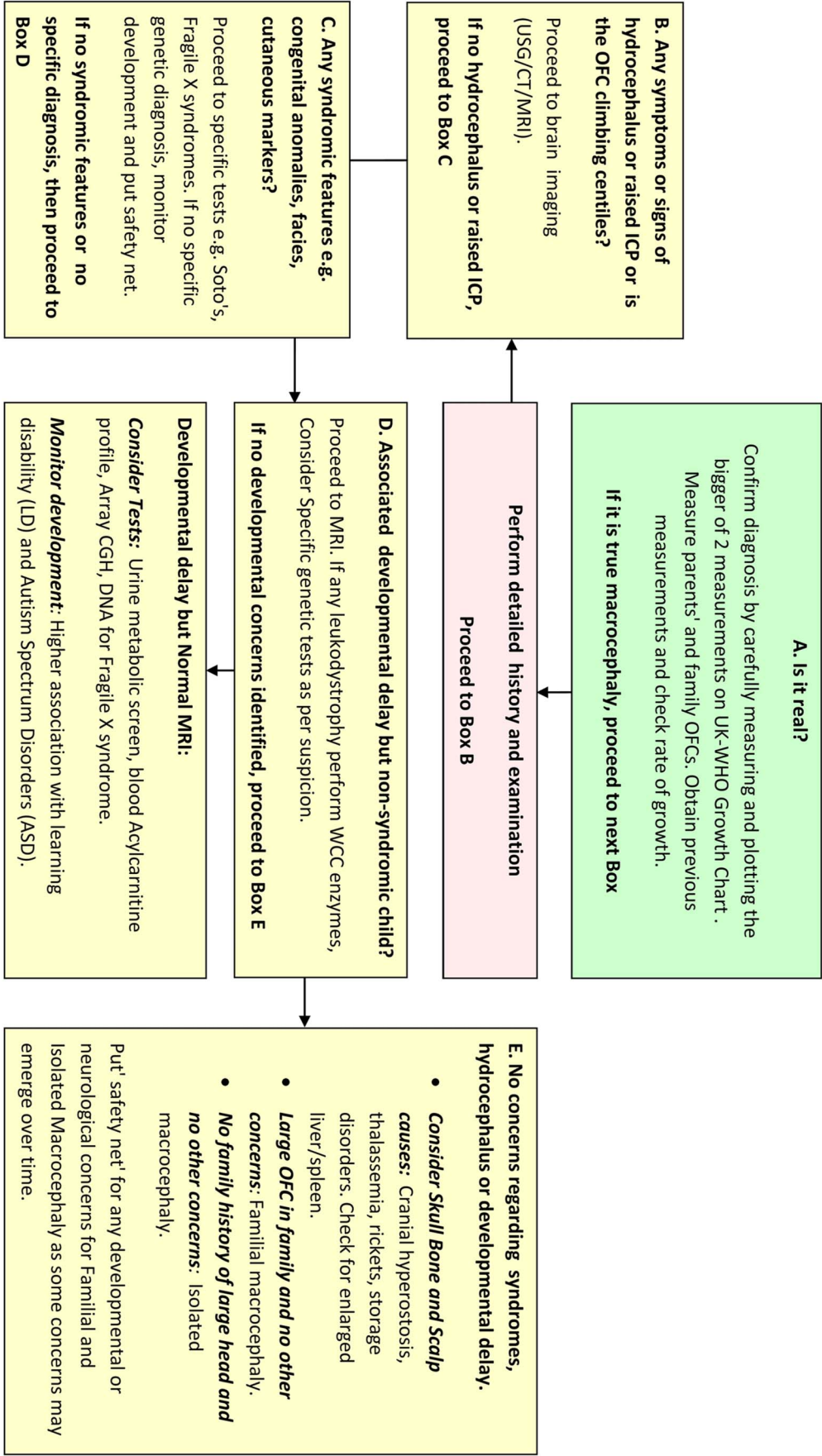


Figure 2 Management of infant with suspected macrocephaly.

indicate a pathological cause as it may occur in benign FM/megalencephaly but should raise concern regarding raised ICP.

3. Are there any high-risk factors that are known to be associated with underlying causes of a large head in infancy? See box 1.
4. Are there any signs or symptoms of raised ICP? See table 1. If there is clinical suspicion of raised ICP, then proceed to neuroimaging, preferably MRI Brain. It is worth noting that all the signs and symptoms outlined are not always present.
5. Is there any history of developmental delay/disorder, seizures, loss of skills, behavioural change or neurological problem in the child? Take a specific developmental history and check developmental levels. Beware of the 'smiley child' who appears 'bright' but is not doing much. Remember, responsive social smile is achieved by 6–8 weeks age!
6. Measure parental and family (sibling, grandparents) head sizes where possible and plot on appropriate charts. The commonest cause for a large head is FM. If the child has no neurological symptoms or signs, has normal development, no signs of raised ICP, has normal systemic examination and the OFC is not climbing centiles, then reassure carers. If the OFC is crossing centiles, it still can be FM, but rule out raised ICP by neuroimaging. DeMyer's criteria for diagnosis of FM are absence of evidence of a syndrome, normal radiographic study of the brain and a parent or sibling with macrocephaly, or macrocephaly that can be traced through several generations.² Discuss the diagnosis with the family and put a 'safety net' of symptoms and signs of raised ICP, seizures, learning delay or developmental concerns for carers to monitor.

7. A thorough examination specifically looking for any known causes of macrocephaly (see figure 1)
 - ▶ dysmorphism
 - ▶ unusual growth patterns, for example, increased growth in Soto's Syndrome
 - ▶ neurocutaneous signs, for example, NF- 1, Tuberous sclerosis
 - ▶ coarse facies or any signs of storage disorders like mucopolysaccharidoses
 - ▶ full neurological examination including checks for focal deficits, co-ordination and fundus examination
 - ▶ signs of raised ICP (see Box 1). Neuroimaging will usually identify the cause of raised ICP, for example, hydrocephalus, intracranial space occupying lesions (expanding cysts or tumours) or subdural collections
 - ▶ hepatosplenomegaly suggesting storage disorders or haematological disorders, for example, thalassemias
- Causes** (see figure 1): The aetiology of macrocephaly can be divided into genetic and non-genetic causes.³
- Management:** Follow the algorithm in figure 2 from A to E to investigate and manage accordingly.

Competing interests None.

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