EVALUATION OF MACROCEPHALY

DEFINITION: OFC greater than 2 standard deviations from the mean or above the 98th percentile

HISTORICAL HIGHLIGHTS:

Congenital vs. Acquired
Static vs. Progressive head growth
Serial head measurements over time are more informative
Premature infants have faster rate of head growth (Use current premature infant growth curves)
Symptoms of increased intracranial pressure
Developmentally normal or delayed
Family History: Familial Megalencephaly/Macrocephaly, Neurocutaneous disorders, Metabolic Disorders

NEUROLOGICAL EXAMINATION

Look for sign of increased intracranial pressure
Measurement of family member’s head circumference
Increased cranial to facial ratio; frontal bossing
Setting-sun sign, CN VI palsies
Long tract signs, i.e. Spasticity, hyperreflexia, upgoing toe to Babinski manuver
Dysmorphic features, neurocutaneous lesions
Height and Weight

CAUSES

THICK SKULL OR SCALP

Anemia, Rickets, Hyperphosphatasemia, Osteogenesis Imperfecta

BENIGN ENLARGEMENT OF THE SUBARACHNOID SPACES (External Hydrocephalus or Benign Subdural Effusions of Infancy)

SUBDURAL FLUID COLLECTIONS
HYDROCEPHALUS

MEGALENCEPHALY

Asymptomatic familial anatomic megalencephaly (see diagnostic criteria)

Metabolic Disorders: Glutaric aciduria type I, Lysosomal diseases, Tay Sachs, Mucopolysaccharidoses

Gigantism: Soto’s syndrome  With skeletal dysplasia: Achondroplasia

Neurocutaneous Disorders: Neurofibromatosis, Tuberous Sclerosis, Hypomelanosis of Ito, Linear sebaceous syndrome

MANAGEMENT

Urgent/Emergent evaluation for progressive macrocephaly and/or signs of intracranial pressure

Neurosurgical consultation

Neuroimaging options:

Head ultrasound for small infants with open fontanels to exclude hydrocephalus/subdural collections expeditiously

Brain CT SCAN: does not require sedation, exposure to radiation

Brain MRI: ideal to evaluate causes of megalencephaly

REFERENCES:
