

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 manueur

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 fontanelles 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:

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Gleason, Dobyns, Palwner and Ashwal. Congenital Structural Defects. In Pediatric Neurology: Principles and Practice, Philadelphia: Mosby Elsevier, 2006