Macrocephaly Etiology

1. with ventricular enlargement

a) (hydrostatic)hydrocephalus (HCP),

● communicating

● obstructive

b) hydranencephaly

c) constitutional ventriculomegaly: ventricular enlargement of no known etiology with normal neurologic function

d) hydrocephalus ex vacuo: loss of cerebral tissue (more often associated with microcephaly, e.g. with TORCH infections)

e) vein of Galen aneurysms:

2. with normal or mildly enlarged ventricles

a) "external hydrocephalus": prominent subarachnoid spaces and basal cisterns; (AKA benign external hydrocephalus)

b) subdural fluid

● hematoma

● hygroma

● effusion benign and symptomatic

● benign subdural collections of infancy

c) cerebral edema: some consider this to be a form of pseudotumor cerebri

● toxic: e.g. lead encephalopathy (from chronic lead poisoning)

● endocrine: hypoparathyroidism, galactosemia, hypophosphatasia, hypervitaminosis A, adrenal insufficiency…

d) familial (hereditary) macrocrania: parents also have large heads, the brains eventually "catch up"

e) idiopathic

f) megalencephaly (AKA macrencephaly): an enlarged brain

g) Neurocutaneous disorders: usually due to increased volume of brain tissue (megalencephaly. Seen especially in neurofibromatosis and congenital hypermelanosis (Ito syndrome). Less common in tuberous sclerosis and Sturge-Weber. Also seen in the rare hemimegalencephaly syndrome

h) arachnoid cyst (AKA subependymal or subarachnoid cyst): a duplication of the ependyma or

arachnoid layer filled with CSF. Usually reach maximal size by 1 month of age and do not enlarge further. Treatment is required in ≈ 30% due to rapid enlargement or growth beyond first month. Cyst may be shunted or fenestrated. Prognosis with true arachnoid cyst is generally good (unlike porencephalic cyst) if no increased ICP or progressive macrocephaly during 1st year of life

i) arteriovenous malformation: especially vein of Galen “aneurysm” . Auscultate for cranial bruit. With vein of Galen aneurysms, macrocephaly may be due to HCP from obstruction of the Sylvian aqueduct. With other malformations, macrocrania may be due to increased pressure in venous system without HCP

j) brain tumors without hydrocephalus: brain tumors are rare in infancy, and most cause obstructive HCP. Tumors that occasionally present without HCP include astrocytomas. May also be seen in the rare diencephalic syndrome, see tumor of anterior hypothalamus

k) “gigantism syndromes”

● Soto’s syndrome: associated with advanced bone age on X-ray, and multiple dysplastic features of face, skin, and bones

● exomphalomacroglossia-gigantism (EMG) syndrome: hypoglycemia (from abnormalities in islets of Langerhans), large birth weight, large umbilicus or umbilical hernia and macroglossia

l) “craniocerebral disproportion”: may be the same as benign extra-axial fluid of infancy

m) achondroplastic dwarf: cranial structures are enlarged but the skull base is small, giving rise to a prominent forehead and an OFC ≥ 97th percentile for age, hypoplasia of midface, and stenosis at foramen magnum. Head growth follows different curve than normal (OFC ≥ 97th percentile for age is not unusual and does not necessitate shunting)

n) Canavan’s disease: AKA spongy degeneration of the brain, an autosomal recessive disease of infancy prevalent among Ashkenazi Jews. Produces symmetrical low attenuation of hemispheric white matter on CT and macrocephaly

o) neurometabolic diseases: usually due to deposition of metabolic substances in the brain. Seen in Tay-Sachs gangliosidosis, Krabbe disease…

3. due to thickening of the skull

a) anemia: e.g. thalassemia

b) skull dysplasia: e.g. osteopetrosis

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