GM1 Gangliosidosis Type 1 (Severe Infantile Type)

(Caffey Pseudo-Hurler Syndrome, Familial Neuorvisceral Lipidosis)

Main Features

- Infancy onset neurodegernation and skeletal abnormalities
- Hypotonia, seizures, dysmorphism, organomegaly
- Rapid psychomotor deterioration starting around 6 months old

Eye Findings

- Cherry-red spot (50%)
- Nystagmus
- Tortuous conjunctival vessels with saccular aneurysms
- Papilledema
- Optic atrophy
- Corneal clouding- rare
- High Myopia

Etiology

- Defect in β-galactosidase-1, all three isoenzymes are deficient (A,B and C)
- Diagnosis confirmed by assay of β-galactosidase in peripheral leukocytes
- Prenatal diagnosis on cultured amniotic fluid cells
- Gene map locus: 3p21.33
- Autosomal recessive

Other Findings

- Facial and peripherial edema
- Kyposis, joint limitation, thick wrists, contractures of elbows and knees, claw hand
- Congestive heart failure, hepatosplenomegaly
- Death usually in early infancy (by age 2)

References

- Pediatric Ophthalmology and Strabismus eds. Wright KW, Spiegel PH. 2nd ed. p. 971,

1039
- OMIM

syndrome