115 年奇美醫院胸腔內科臨床病例討論會

1 時間：115 年 06 月 09日 PM: 4:00-5:00
2 課程活動題目:Niemann-Pick disease in CXR
3 主講人：柯獻欽
4 地點： 奇美醫學中心 10 樓空橋討論室
5 聯絡人：黎安騏（06-2812811 #57132）
6 摘要：

Niemann-Pick A and B
AR, chrom 11p15, deficiency in sphingomyelinase (A Ashkenazi Jews, B Mediterranean)
Type A (infantile)
Insidious onset FTT, hepatomegaly predominates, splenomegaly, pulmonary dz.
Loss of visual contact, developmental regression, sz
Hypotonia, decreased DTRs, supranuclear gaze palsy
Neuronal storage- cytoplasmic swelling, atrophy of cerebellum
Death in 1st few yrs
"Cherry red spot" nl macula in middle of retina changing to gray from storage
Bone marrow with foam cells (Niemann-Pick cells)
CXR mottled pattern (like miliary TB)

Type B
Hepatomegaly-liver dz, splenomegaly-pancytopenia, pulmonary macrophage infiltration
Usual no neurological problems but may develop ataxia, macular changes, nl lifespan

Dx: measure enzyme activity in leukocytes/fibroblasts
Tx: none

Niemann-Pick Type C and D (Nova Scotia variant)
Defect in cholesterol esterification leading to lysosomal accumulation
Swelling of proximal neurites in cortex (like Batten dz & gangliosidoses), NFTs (like AD)
May present early with neonatal conjugated hyperbilirubinemia or later
Developmental delay then regression, wide range of cognitive problems
Sz, dystonia, ataxia, supranuclear gaze palsy, cataplexy

Hepatic failure, death in 2nd decade
Dx: stain cultured fibroblasts with filipin for cholesterol, then check esterification
Tx: none, prognosis based on age of onset

Symptoms:
Type A Niemann-Pick begins in the first few months of life. Symptoms may include:

a large abdomen within 3 to 6 months
liver or spleen enlargement
cherry red spot in the eye
feeding difficulties
progressive loss of early motor skills
(generally) a very rapid decline leading to death by two to three years of age

Type B is biochemically similar to Type A but the symptoms are more variable. Neurological involvement, such as loss of motor skills, is slight to none. Common symptoms usually appear in infancy or childhood. Progression of Type B is generally much slower than with Type A and many people live into adulthood.

liver or spleen enlargement
cherry red spot in the eye
shortness of breath (may require oxygen)
repeated lung infections

Type C Niemann-Pick usually affects children of school age, but the disease may strike at any time from early infancy to adulthood. Symptoms may include:

jaundice at (or shortly after) birth
an enlarged spleen and/or liver
difficulty with upward and downward eye movements (Vertical Supranuclear Gaze Palsy).
unsteadiness of gait, clumsiness, problems in walking ("ataxia")
difficulty in posturing of limbs ("dystonia")
slurred, irregular speech ("dysarthria")
learning difficulties and progressive intellectual decline ("dementia")
sudden loss of muscle tone which may lead to falls ("cataplexy")
tremors accompanying movement and, in some cases, seizures
psychiatric problems of unknown cause in teens and adults

Type C is the most variable form of the disease. Symptoms may appear and then disappear. Some symptoms may never appear. The rate the disease progresses is different from person to person. The rate of progress for an individual will change over time.

Type C is often incorrectly diagnosed. Some of the common errors are:

Attention Deficit Disorder (ADD)
Learning Disability
Retardation
Delayed Development

Vertical Supranuclear Gaze Palsy (VSGP or VGP) is highly suggestive of Type C. VSGP is the inability to move the eyes up and down. Parents often notice this when their child walks up and down stairs, watches TV while sitting on the floor, or in similar situations - the child tilts their head to see instead of moving their eyes. Liver or spleen problems in the first few months after birth are also highly suggestive of Type C.