9. ACEM-Pathsoc Manchester 06.07.2006

Lafora Body Disease presentation
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Extracted from presentation by Bart Wagner

Clinical details:
16-year-old female
Presented with Grand mal tonic/clonic seizures
Later developed absence, visual and myoclonic seizures
Developmental delay and progressive mental deterioration
Examination revealed cerebellar ataxia and muscle wasting with myoclonus

Deep skin biopsy from axilla for histology and electron microscopy taken.

Histology showed both eccrine and apocrine sweat glands – apocrine glands are deeper, longer and of larger diameter than eccrine
PAS stain reveals inclusions within myoepithelial cells in apocrine glands and not in eccrine glands – varying between 5 and none per tubular profile.
EM showed inclusions approximately half the size of a nucleus composed of granular and irregular filamentous material approximately 6nm in diameter. These inclusions are not membrane bound.
At post mortem Lafora bodies were demonstrated by PAS stain. They were found in the neurones of the dentate fascia in the cerebellum.

Practice points:
PAS false positive granules can be seen in eccrine and apocrine glands but they are found in the epithelial cells not the myoepithelial cells.
Lafora bodies are said to be seen in eccrine glands if taken away from area in which apocrine glands are founds otherwise not.

References:
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