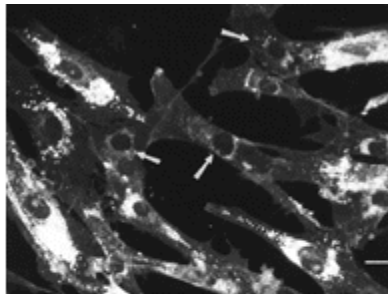




Niemann-Pick disease



Cells stained to show unesterified cholesterol in NP-C cells (white). The arrows show cell normalized by transfection with NPC1 DNA. [Reproduced with permission from Carstea et al. (1997) *Science* 277, 228-231.]

In 1914, German pediatrician Albert Niemann described a young child with brain and nervous system impairment. Later, in the 1920's, Luddwick Pick studied tissues after the death of such children and provided evidence of a new disorder, distinct from those storage disorders previously described.

Today, there are three separate diseases that carry the name Niemann-Pick: Type A is the acute infantile form, Type B is a less common, chronic, non-neurological form, while Type C is a biochemically and genetically distinct form of the disease. Recently, the major locus responsible for Niemann-Pick type C (NP-C) was cloned from chromosome 18, and found to be similar to proteins that play a role in cholesterol homeostasis.

Usually, cellular cholesterol is imported into lysosomes—'bags of enzymes' in the cell—for processing, after which it is released. Cells taken from NP-C patients have been shown to be defective in releasing cholesterol from lysosomes. This leads to an excessive build-up of cholesterol inside lysosomes, causing processing errors. NPC1 was found to have known sterol-sensing regions similar to those in other proteins, which suggests it plays a role in regulating cholesterol traffic.

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