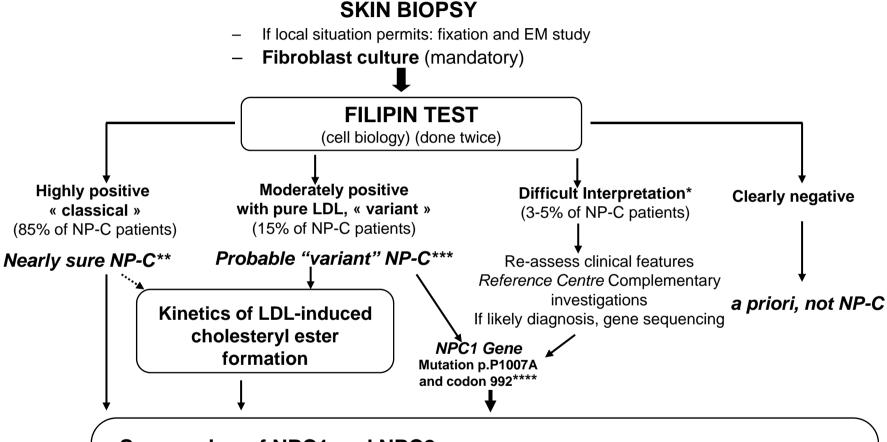
- Bone marrow (useful, not mandatory): May show foam cells (filipin + if tested for this stain)
- If a liver biopsy is performed for cholestatic liver disease, fixation for EM study is essential
- Serum chitotriosidase: useful, not mandatory; generally (not always) elevated activity
- Isolated (hepato)splenomegaly: enzymatic exclusion of Gaucher and Niemann-Pick B = prerequisite
- Provide the laboratory with sufficient clinical data (essential for correct interpretation of the results)



Sequencing of NPC1 and NPC2 genes

- Depending on countries, study first *NPC1* p. I1061T or other most prevalent common mutation
- Conclude quickly on NPC2 if child < 8-10 months
- NPC1: numerous polymorphisms!!! check allele segregation from parental study
- often need to study both gDNA and cDNA