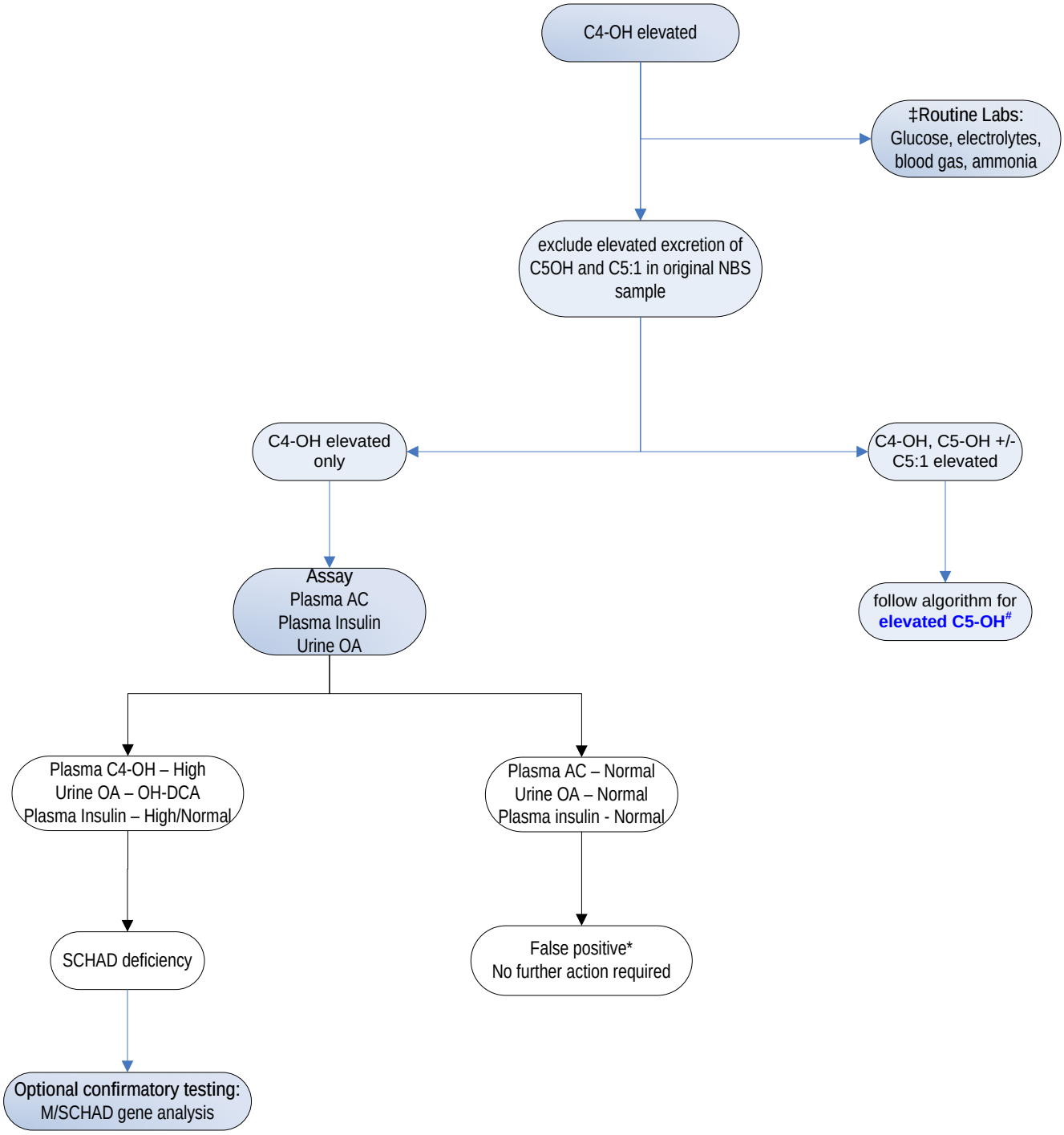
C4-OH elevated
(screening lab not measuring C5-OH and C5:1)



Abbreviations:
AC = acylcarnitines
C4-OH – C4-hydroxy acylcarnitine
C5-OH – C5-hydroxy acylcarnitine
C5:1 – tiglylcarnitine
DCA – dicarboxylic acids
OA – organic acids
SCHAD – short-chain 3-hydroxy acyl-CoA dehydrogenase

‡ = When the positive predictive value of screening is sufficiently high and the risk to the infant is high, some initiate diagnostic studies that are locally available at the same time as confirmation of the screening result is done.

* = M/SCHAD is a secondary target due to limited understanding of the condition. Metabolic disease specialists should be consulted and may consider enzyme assays in fibroblasts and/or HAD1 mutation analysis despite normal plasma AC, urine OA, and plasma insulin.

= http://www.acmg.net/resources/policies/ACT/Visio-C5-OH(4-29-06).pdf

(Funded in part through MCHB/HRSA/HHS grant #U22MC03957)