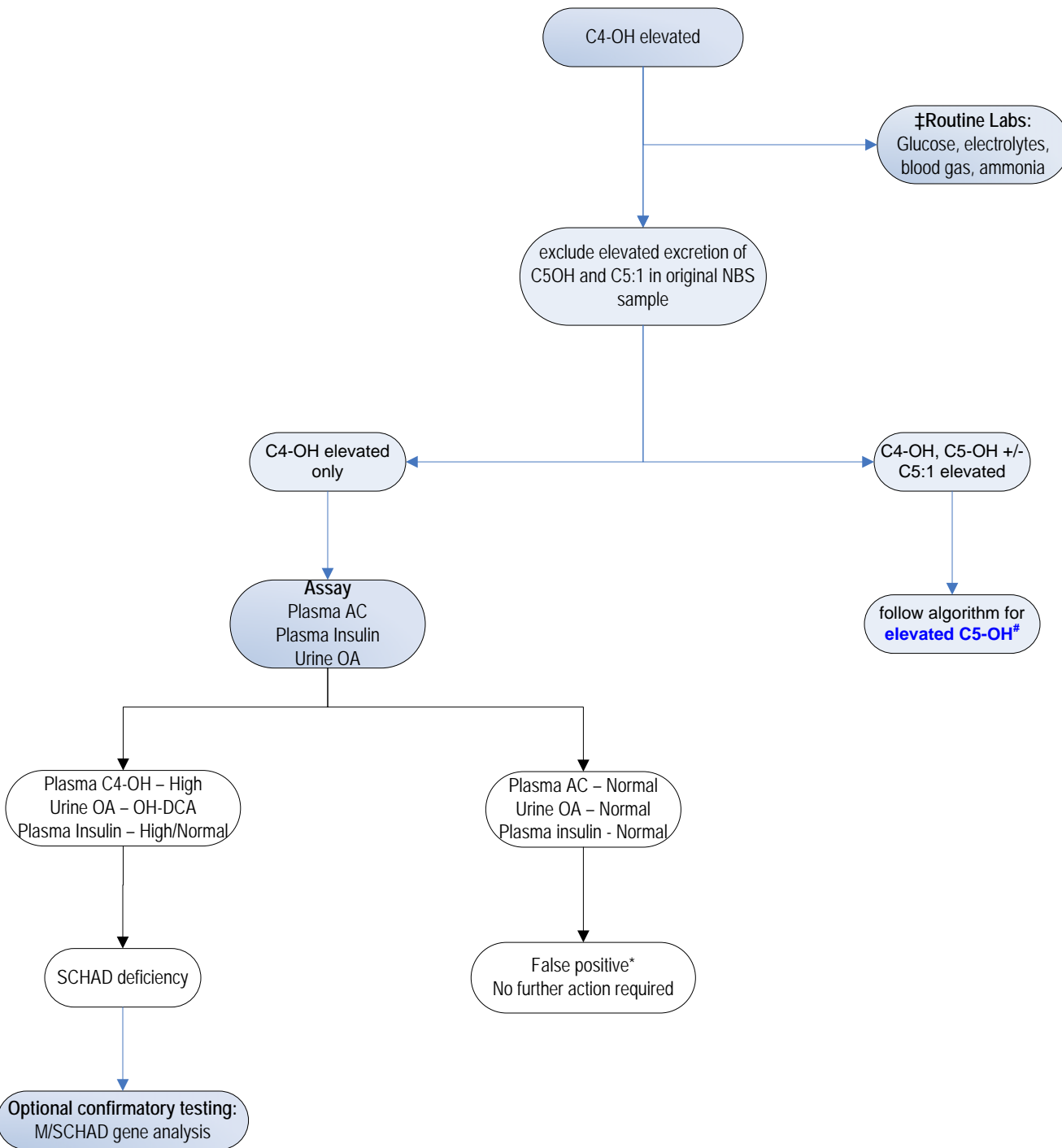




C4-OH elevated (screening lab not measuring C5-OH and C5:1)



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

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.