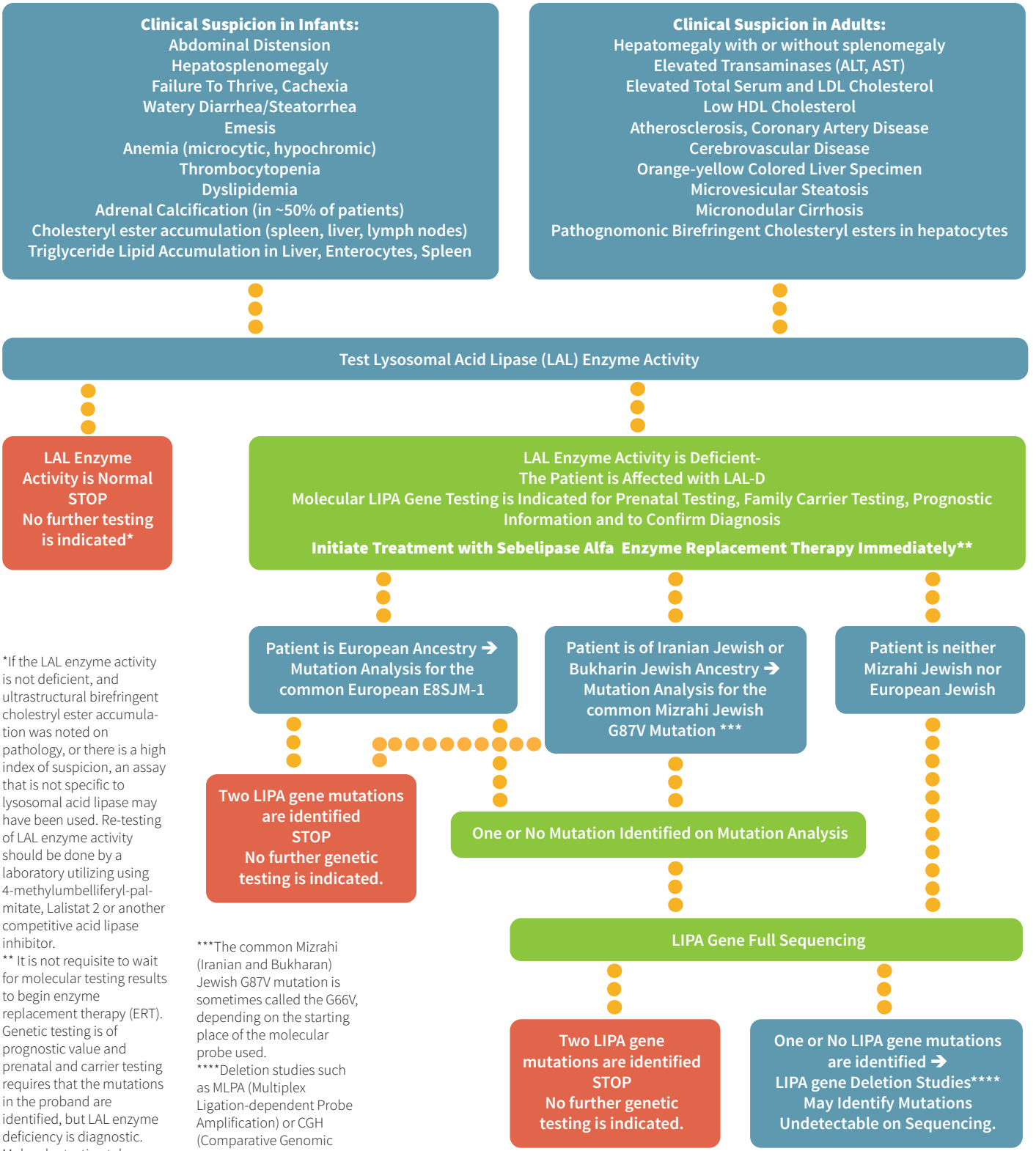
Diagnostic Testing Algorithm
Lysosomal Acid Lipase Deficiency



*If the LAL enzyme activity is not deficient, and ultrastructural birefringent cholestryl ester accumulation was noted on pathology, or there is a high index of suspicion, an assay that is not specific to lysosomal acid lipase may have been used. Re-testing of LAL enzyme activity should be done by a laboratory utilizing using 4-methylumbelliferyl-palmitate, Lalistat 2 or another competitive acid lipase inhibitor.

** It is not requisite to wait for molecular testing results to begin enzyme replacement therapy (ERT). Genetic testing is of prognostic value and prenatal and carrier testing requires that the mutations in the proband are identified, but LAL enzyme deficiency is diagnostic. Molecular testing takes several weeks. In severe infantile LAL-D any delay in treatment may result in mortality.

***The common Mizrahi (Iranian and Bukharan) Jewish G87V mutation is sometimes called the G66V, depending on the starting place of the molecular probe used.

****Deletion studies such as MLPA (Multiplex Ligation-dependent Probe Amplification) or CGH (Comparative Genomic Hybridization) may identify mutations, such as deletions that would not be detectable using sequencing methods.