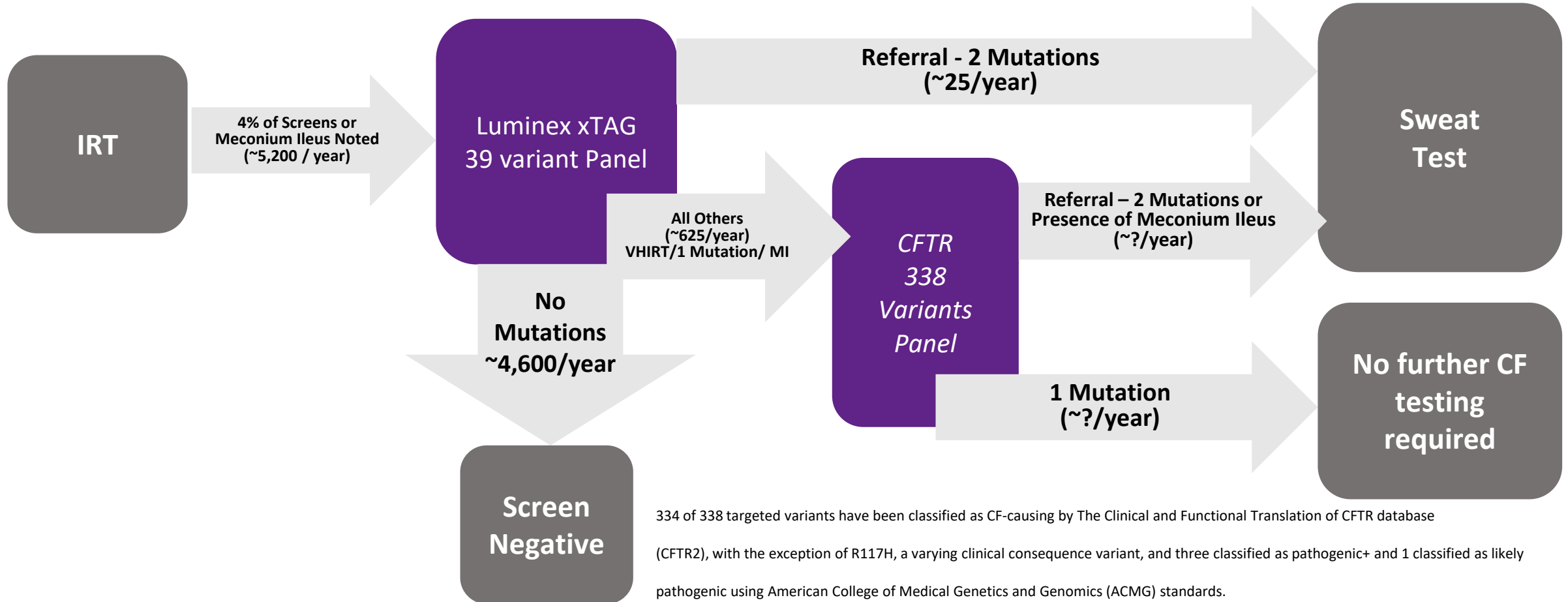


Cystic Fibrosis

NBS Testing Algorithm for the Pennsylvania Department of Health

Last Updated: September 21, 2022



334 of 338 targeted variants have been classified as CF-causing by The Clinical and Functional Translation of CFTR database (CFTR2), with the exception of R117H, a varying clinical consequence variant, and three classified as pathogenic+ and 1 classified as likely pathogenic using American College of Medical Genetics and Genomics (ACMG) standards.

Large deletion/duplications defined as CF-causing by CFTR are not included. This follows other state algorithms.