Canadian College of Medical Geneticists - CF Testing/Screening Guidelines

Revised by the CCMG Molecular Genetics Committee

Approved by the CCMG Board of Directors January 20, 2011.

1. The CCMG endorses that CFTR testing may be indicated for individuals or families who may be at increased risk of CF or a CFTR-related disorder due to considerations of family history or clinical manifestations and support the offer of genetic testing for the following indications:

   • confirmation of a diagnosis of cystic fibrosis or a CFTR-related disorder
   • carrier testing in individuals with a positive family history of CF or a CFTR-related disorder
   • carrier testing in partners of individuals with a positive family history of CF or a CFTR-related disorder
   • prenatal diagnosis for pregnancies at 25% or greater risk of CF
   • prenatal diagnosis for pregnancies in which a diagnosis of echogenic bowel has been identified in the fetus
   • newborn screening with screen positive results for cystic fibrosis

2. At this time, the Board does not endorse carrier screening of the general adult population.