The ADMINISTRATIVE Policy Statement detailed above has received due consideration as defined in the

- Autosomal Recessive – A trait or disorder requiring a deleterious variant in both copies of the gene to express a phenotype.
- Carrier – An individual with a gene variant for a disease or disorder who can pass the variant on to offspring but does not have symptoms or features of the disorder.
- Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) – Transmembrane protein that functions as a regulated anion channel to maintain a gradient necessary for normal cell function. Mutations that alter the function of the CFTR protein are associated with CF.
- CFTR-Related Disorder – A single pathogenic CFTR variant leading to the development of disease limited to one organ system in an individual who does not fit the CF diagnosis.
- CFTR-Related Metabolic Syndrome (CRMS) – Infants and children who are asymptomatic but have positive CF screening results. Found in 3-4% of infants with a positive newborn CF screen, CRMS is also known as CF screen positive, inconclusive diagnosis (CFSPID).
- Immunoreactive Trypsinogen (IRT) – A pancreatic enzyme precursor measured in newborns to screen for the presence of CF.

D. Policy

I. Genetic testing for CF should only be performed once in a lifetime and the results documented in the member's health record. All genetic testing for CF should use currently recommended ACMG CFTR panels. Prior authorization is required for all CF genetic tests.

II. Diagnostic testing is considered medically necessary when the member meets any of the following criteria:
   A. clinical presentation of CF
   B. infertility from oligospermia/azoospermia/congenital bilateral absence of vas deferens (CBAVD)
   C. infant with meconium ileus or other symptoms indicative of CF but unable to produce adequate amounts of sweat for a sweat c9y998t(e)-ent ev.004 (t)-4.004 (e a)1998 ( )-4

A. repeat testing
B. fetal testing
C. gene sequencing of

9. Deignan JL, Gregg AR, Grody WW, et al. Updated recommendations for CFTR carrier screening: a position statement of the American College of Medical Genetics and Genomics (Acs 61, 44 86 (li)612 792 re A126.02 682.005 ( )-4.00.004 (Ai)5 (cs)5 (.996 ( )5.996