

	Cystic Fibrosis	CFTR-Related Disease		Infertility
Test Criteria	<ul style="list-style-type: none"> · Clinical diagnosis <ul style="list-style-type: none"> · Elevated sweat chloride (≥ 60 mmol/L) · Borderline sweat chloride (30-60 mmol/L) · Positive newborns screen · Meconium ileus 	<p style="text-align: center;">Pulmonary</p> <ul style="list-style-type: none"> · Bronchiectasis · Recurrent pseudomonas infections 	<p style="text-align: center;">Pancreatitis</p> <ul style="list-style-type: none"> · Idiopathic sporadic pancreatitis <30 y.o · Familial pancreatitis (≥ 2 family members) 	<ul style="list-style-type: none"> · C(B)AVD · Azoospermia · Oligospermia
Exclusions	Isolated aquagenic wrinkling of the palms	Isolated aquagenic wrinkling of the palms		Infertility NOS
Testing Algorithm	<p>For patients with a clinical diagnosis</p> <ul style="list-style-type: none"> · CF 39 recurrent variant panel · Reflex to sequencing and MLPA, if indicated <p>Newborn screen</p> <ul style="list-style-type: none"> · Targeted testing for the NBS variants (please provide) <p>For borderline sweat chloride, meconium ileus</p> <ul style="list-style-type: none"> · CF 39 recurrent variant panel · If one variant identified-> reflex to sequencing and MLPA, if requested · If no variant identified-> no further testing 	<ul style="list-style-type: none"> · See algorithm for borderline sweat chloride 	<ul style="list-style-type: none"> · See algorithm for borderline sweat chloride 	<ul style="list-style-type: none"> · CF 39 recurrent variant panel · For C(B)AVD <ul style="list-style-type: none"> · Reflex to sequencing and MLPA when requested · For azoo/oligospermia <ul style="list-style-type: none"> · <u>If one variant or ST is identified</u> -> reflex to sequencing and MLPA, if requested · <u>If no variant identified</u> -> no further testing

	Parental Testing	Fetal Testing		
Test Criteria	<ul style="list-style-type: none"> · Echogenic bowel on ultrasound · Dilated bowel · Fetal ascites 	<ul style="list-style-type: none"> · Both parents known to be carriers 	<ul style="list-style-type: none"> · Echogenic bowel on ultrasound AND · One parent is a known carrier, other parent cannot be tested 	<ul style="list-style-type: none"> · Echogenic bowel on ultrasound AND · One parent negative, other parent cannot be tested
Exclusion				
Testing algorithm	<ul style="list-style-type: none"> · CF 39 recurrent variant panel for both parents in parallel (regardless of ethnicity) <u>If one parent found to be a carrier</u> · Will receive a call from the laboratory. Reflex to sequencing and MLPA of the other parent upon request <u>If none of the parents are positive by the 39 recurrent variant panel</u> · No further testing, regardless of ethnicity 	<ul style="list-style-type: none"> · Testing of the familial variants only (please provide) 	<ul style="list-style-type: none"> · CF 39 recurrent variant panel · Reflex to sequencing and MLPA when requested 	<ul style="list-style-type: none"> · CF 39 recurrent variant panel · Reflex to sequencing and MLPA when requested

	Positive Family History, variants known	Positive Family History, variants unknown	Partner is affected, or is a known carrier	Population Screening
Test Criteria	<ul style="list-style-type: none"> Known familial variant(s) (please provide the report and a positive control, if available) 	<ul style="list-style-type: none"> Reported positive family history (report unavailable) 	<ul style="list-style-type: none"> Partner is a known carrier Partner is affected (has two pathogenic variants in trans) 	<ul style="list-style-type: none"> French Canadians with family origins from the Quebec regions of Saguenay Lac-St-Jean and Charlevoix (please specify)
Exclusion	<ul style="list-style-type: none"> Familial variant is 5T, unless the 5T has been reported with a TG tract and this is also requested 		<ul style="list-style-type: none"> Partner is a 5T carrier only 	<ul style="list-style-type: none"> Other ethnicity-based population screening Egg/sperm donors (unless otherwise indicated)
Testing algorithm	<ul style="list-style-type: none"> Targeted testing for the familial variants 	<ul style="list-style-type: none"> CF 39 recurrent variant panel 	<p>Partner is a known carrier:</p> <ul style="list-style-type: none"> CF 39 recurrent variant panel <p>Partner has two pathogenic variants, in trans:</p> <ul style="list-style-type: none"> CF 39 recurrent variant panel Reflex to sequencing and MLPA, if requested 	<ul style="list-style-type: none"> CF 39 recurrent variant panel