Fragile X Syndrome, Carrier

Order Name: **FRAGILE X**
Test Number: 0117075
Revision Date: 03/11/2024

TEST NAME	METHODOLOGY	LOINC CODE
Fragile X Syndrome, Carrier	PCR/Capillary Electrophoresis	53039-4

SPECIMEN REQUIREMENTS

Specimen	Specimen Volume (min)	Specimen Type	Specimen Container	Transport Environment
Preferred	**8.5 mL (3 mL)**	**Whole Blood**	**EDTA (Lavender Top)**	**Room Temperature**
Alternate 1	**1**	**See Instructions**	**See Instructions**	**Room Temperature**

Instructions

Specimen Type: 8.5 mL whole blood from Lavender-top (EDTA) tube OR PurFlock buccal swab kit or Oragene Dx saliva kit
Collection: Standard phlebotomy. Follow PurFlock buccal swab kit or Oragene Dx 500 saliva kit collection instructions. Do not eat, drink, smoke or chew gum 30 minutes prior to collection.
Specimen Storage: Room temperature
Stability: Whole blood: 14 days at room temperature or 30 days at 4'C

GENERAL INFORMATION

Expected TAT	6-14 Days
Clinical Use	This test is used for carrier screening for fragile X syndrome (FMR1). ?Limitations: Technologies used do not detect germline mosaicism and do not rule out the presence of large chromosomal aberrations including rearrangements and gene fusions, or variants in regions or genes not included in this test, or possible inter/intragenic interactions between variants, or repeat expansions. Variant classification and/or interpretation may change with time if more information becomes available. False positive or false negative results may occur for reasons that include: rare genetic variants, sex chromosome abnormalities, pseudogene interference, blood transfusions, bone marrow transplantation, somatic or tissue-specific mosaicism, mislabeled samples or erroneous representation of family relationships. This test was developed and its performance characteristics determined by Labcorp. It has not been cleared or approved by the Food and Drug Administration.
Performing Labcorp Test Code	481684
Notes	Polymerase chain reaction (PCR) followed by capillary electrophoresis, with reflex to AGG interruption analysis and methylation PCR analysis for positive samples
CPT Code(s)	81243
Lab Section	Reference Lab