TECHNICAL BULLETIN

DISCONTINUED TEST

NOTIFICATION DATE: August 13, 2024

EFFECTIVE DATE: Immediate

Inherigen and Ashkenazi Jewish Carrier Screening Tests

On the effective date, Bioreference Laboratories will discontinue several of their pan- ethnic and Ashkenazi Jewish reproductive carrier screening tests and panels. Northwell Health Laboratories has several replacement options performed by Natera Laboratories. Below is a table detailing the test requirements, along with an associated table that outlines the corresponding Natera Horizon carrier screen tests, which include the genes found in the current Bioreference tests that will be discontinued.

The Horizon Carrier Screen tests offered by Natera Laboratories, are genetic screening tests designed to identify carriers of specific inherited genetic disorders. These tests utilize next-generation sequencing (NGS) methodology to detect mutations in genes associated with a wide range of genetic conditions including cystic fibrosis, spinal muscular atrophy, and fragile X syndrome. The Horizon carrier screen tests can cover a comprehensive panel of up to 421 conditions, providing detailed insights into an individual's genetic makeup. The primary purpose of these tests is to inform prospective parents about their carrier status, helping them make informed decisions regarding family planning and reproductive health. Natera's Horizon tests are known for their accuracy, extensive coverage, and the ability to deliver actionable information that can guide clinical decisions and personalized care strategies.

Test	Horizon Carrier Screen	Bioreference Carrier Screen Tests		
Requirement/Parameters	Tests (New)	(Previous)		
Method:	Next Generation Sequencing (NGS), Polymerase Chain Reaction (PCR)	Polymerase Chain Reaction (PCR)		
Specimen Requirements:	Whole Blood	Whole Blood		
Submission Container/Tube:	Lavender top or Buccal Swab	Lavender top or Buccal Swab		
Specimen Volume/Minimum Volume:	10 ml/4 mL	4 mL		
Collection Instructions:	Whole Blood	Whole Blood		
Specimen Stability:	72 Hours Room Temperature 7 Days Refrigerated Buccal Swab- 6 Days Room Temperature	72 Hours Room Temperature 7 Days Refrigerated Buccal Swab- 6 Days Room Temperature		
Reference Range:	Negative	Negative		

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Test Ordering Crosswalk					
	Horizon Carrier Screen Tests (New) Bioreference Carrier Screen Tests (Page 1997)				
Test Name	Horizon 274 or Horizon 421	Inherigen Carrier Screening			
Test Order	HORIZON274 or HORIZON421	INHERIGEN or INHERIGENPLUS			
Computer Interface Code	PDM# 225281 or 225043	PDM# 1659837 or 1659848			
Test Name	Horizon 106*	Ashkenazi Jewish 18 Panel	PDM # 1659843		
	*Electronic Test order currently	Ashkenazi Jewish 25 Panel	PDM # 1659849		
	unavailable, please utilize Horizon 274	Bloom Syndrome	PDM # 5907077		
	until test is offered electronically	Maple Syrup Urine Disease	PDM # 5907084		
		Nemaline Myopathy 2	PDM # 5909100		
		Mucolipidosis Type IV	PDM # 5910090		
		Niemann-pick Type A+B	PDM # 5907079		
		Usher Syndrome Type 1F	PDM # 5909105		
		Usher Syndrome Type 3	PDM # 5909110		
		Fanconi Anemia, Type C	PDM # 5907081		
		Glycogen Storage Type 1A	PDM # 5907078		
		Dihydrolipomide Dehydrogenase Deficiency	PDM # 5909120		
Test Order	HORIZON106*	See test directory			
Computer Interface Code	PDM# 245346	See above			
Test Name	Horizon 14	Tay-Sachs DNA	PDM # 5900774		
		Canavan Disease	PDM # 5910540		
		Familial Dysautonomia	PDM # 5900776		
		Gaucher Carrier Screen	PDM # 5910530		
Test Order	HORIZON14	See test directory			
Computer Interface Code	PDM# 245600	See above			