10/21/2014 - New Release, Quest Diagnostics Nichols Institute, Valencia

Revision Message!
Please note: 10/29/14 communication revision as all tests are available for New York patient testing.

NEW TESTS Please Note: Not all test codes assigned to each assay are listed in the table of contents. Please refer to the complete listing on the page numbers indicated.			
Test Code	**Test Name**	**Effective Date**	**Page #**
92587	**BRCAvantage™ Plus *(BRCA1, BRCA2, TP53, PTEN, CDH1, STK11, PALB2)***	11/3/2014	1
92573	**BRCAvantage™ with Reflex to Breast Plus Panel**	11/3/2014	3
92586	**Breast Plus Panel without BRCA *(TP53, PTEN, CDH1, STK11, PALB2)***	11/3/2014	5
92568	**CDH1 Sequencing and Deletion/Duplication**	11/3/2014	6
92571	**PALB2 Sequencing and Deletion/Duplication**	11/3/2014	7
92566	**PTEN Sequencing and Deletion/Duplication**	11/3/2014	8
92565	**STK11 Sequencing and Deletion/Duplication**	11/3/2014	9
92560	**TP53 Sequencing and Deletion/Duplication**	11/3/2014	9

New Test Offerings

The following tests will be available through Quest Diagnostics on the dates indicated below.

BRCAvantage™ Plus *(BRCA1, BRCA2, TP53, PTEN, CDH1, STK11, PALB2)*	
Revision Message!	**Please note: The message was updated to remove the New York Patient Testing restriction effective 10/29/14.**
Clinical Significance	**10% of breast cancer cases that are hereditary are caused by a few genes. BRCA1 and BRCA2 account for most (~80%) of hereditary breast cancers. Five other medium risk genes include CDH1, PALB2, PTEN, STK11 and TP53. Sequence analysis and deletion/duplication analysis of these five genes can complement BRCA testing and further elucidate an individual's hereditary breast cancer risk.**
Effective Date	**11/3/2014**
Test Code	**92587**
CPT Codes	**81211, 81213, 81405 (x 2), 81479, 81321, 81323, 81406 (x 2), 81404**
Specimen Requirements	**4 mL (2 mL minimum) whole blood collected in an EDTA (lavender-top) tube**
Reject Criteria	**Gross hemolysis; clotted specimens**
Instructions	**Whole Blood: Normal phlebotomy procedure; specimen stability is crucial. Store and ship room temperature immediately. Do not freeze. Send report of results for family member with known BRCA mutation.**
Transport Temperature	**Room temperature**
Specimen Stability	**Room temperature and Refrigerated: 8 days** **Frozen: Unacceptable**
Set-up/Analytic Time	**Set up: Tues, Thurs, Sat; Report available: 14 days from completed pre-authorization**
Reference Range	**Accompanies report**
Methodology	**Exon Capture, Next Generation Sequencing, Multiplex Ligation-dependent Probe Amplification**

Performing Site	**Quest Diagnostics Nichols Institute, San Juan Capistrano**

Interface Mapping

92587-1-Interpretation Summary		
Result Code	**Type**	**Result Name**
86007572	**Prompt-Result (no return)**	**Ethnicity:**
86011589		**Interpretation Summary**

92587-2-BRCA1 Sequencing and Del/Dup	
Result Code	**Result Name**
86010069	**BRCA1 Sequencing**
86010070	**BRCA1 Seq Interp**
86010071	**BRCA1 Del/Dup**
86010072	**BRCA1 Del/Dup Interp**

92587-3-BRCA2 Sequencing and Del/Dup	
Result Code	**Result Name**
86010073	**BRCA2 Sequencing**
86010074	**BRCA2 Seq Interp**
86010075	**BRCA2 Del/Dup**
86010076	**BRCA2 Del/Dup Interp**

92587-4-TP53 Sequencing and Del/Dup	
Result Code	**Result Name**
86011410	**TP53 Sequencing**
86011411	**TP53 Seq Interp**
86011412	**TP53 Del/Dup**
86011413	**TP53 Del/Dup Interp**

92587-5-PTEN Sequencing and Del/Dup	
Result Code	**Result Name**
86011520	**PTEN Sequencing**
86011521	**PTEN Seq Interp**
86011522	**PTEN Del/Dup**
86011523	**PTEN Del/Dup Interp**

92587-6-CDH1 Sequencing and Del/Dup	
Result Code	**Result Name**
86011527	**CDH1 Sequencing**
86011528	**CDH1 Seq Interp**
86011529	**CDH1 Del/Dup**
86011530	**CDH1 Del/Dup Interp**

92587-7-STK11 Sequencing and Del/ Dup	
Result Code	**Result Name**
86011513	**STK11 Sequencing**
86011514	**STK11 Seq Interp**
86011515	**STK11 Del/Dup**
86011516	**STK11 Del/Dup Interp**
92587-8-PALB2 Sequencing and Del/Dup	
Result Code	**Result Name**
86011534	**PALB2 Sequencing**
86011535	**PALB2 Seq Interp**
86011536	**PALB2 Del/Dup**
86011537	**PALB2 Del/Dup Interp**
92587-9-Comprehensive Report and Additional Information	
Result Code	**Result Name**
86011590	**Comprehensive Interp**
86011591	**Additional Information**

BRCAvantage™ with Reflex to Breast Plus Panel	
Revision Message!	**Please note: The message was updated to remove the New York Patient Testing restriction effective 10/29/14.**
Clinical Significance	**10% of breast cancer cases that are hereditary are caused by a few genes. BRCA1 and BRCA2 account for most (~80%) of hereditary breast cancers. Five other medium risk genes include CDH1, PALB2, PTEN, STK11 and TP53. Sequence analysis and deletion/duplication analysis of these five genes can complement BRCA testing and further elucidate an individual's hereditary breast cancer risk.**
Effective Date	**11/3/2014**
Test Code	**92573**
CPT Codes	**81211, 81213**
Specimen Requirements	**4 mL (2 mL minimum) whole blood collected in an EDTA (lavender-top) tube**
Reject Criteria	**Gross hemolysis; clotted specimens**
Instructions	**Whole Blood: Normal phlebotomy procedure; specimen stability is crucial. Store and ship room temperature immediately. Do not freeze. Send report of results for family member with known BRCA mutation.**
Transport Temperature	**Room temperature**
Specimen Stability	**Room temperature and Refrigerated: 8 days** **Frozen: Unacceptable**
Set-up/Analytic Time	**Set up: Tues, Thurs, Sat; Report available: 14 days from completed pre-authorization**
Reference Range	**Accompanies report**
Methodology	**Exon Capture, Next Generation Sequencing, Multiplex Ligation-dependent Probe Amplification**
Performing Site	**Quest Diagnostics Nichols Institute, San Juan Capistrano**

Interface Mapping

92573-1-Interpretation Summary		
Result Code	**Type**	**Result Name**
86007572	Prompt-Result (no return)	Ethnicity:
86011541		Interpretation Summary

92573-2-BRCA1 Sequencing and Del/Dup	
Result Code	**Result Name**
86010069	BRCA1 Sequencing
86010070	BRCA1 Seq Interp
86010071	BRCA1 Del/Dup
86010072	BRCA1 Del/Dup Interp

92573-3-BRCA2 Sequencing and Del/Dup	
Result Code	**Result Name**
86010073	BRCA2 Sequencing
86010074	BRCA2 Seq Interp
86010075	BRCA2 Del/Dup
86010076	BRCA2 Del/Dup Interp

** TR 92573-4-TP53 Sequencing and Del/Dup*	
Result Code	**Result Name**
86011410	TP53 Sequencing
86011411	TP53 Seq Interp
86011412	TP53 Del/Dup
86011413	TP53 Del/Dup Interp

**TR 92573-5-PTEN Sequencing and Del/Dup*	
Result Code	**Result Name**
86011520	PTEN Sequencing
86011521	PTEN Seq Interp
86011522	PTEN Del/Dup
86011523	PTEN Del/Dup Interp

**TR 92573-6-CDH1 Sequencing and Del/Dup*	
Result Code	**Result Name**
86011527	CDH1 Sequencing
86011528	CDH1 Seq Interp
86011529	CDH1 Del/Dup
86011530	CDH1 Del/Dup Interp

**TR 92573-7-STK11 Sequencing and Del/Dup*

	<table><tr><th>Result Code</th><th>Result Name</th></tr><tr><td>86011513</td><td>STK11 Sequencing</td></tr><tr><td>86011514</td><td>STK11 Seq Interp</td></tr><tr><td>86011515</td><td>STK11 Del/Dup</td></tr><tr><td>86011516</td><td>STK11 Del/Dup Interp</td></tr><tr><td colspan="2">*TR 92573-8-PALB2 Sequencing and Del/Dup</td></tr><tr><th>Result Code</th><th>Result Name</th></tr><tr><td>86011534</td><td>PALB2 Sequencing</td></tr><tr><td>86011535</td><td>PALB2 Seq Interp</td></tr><tr><td>86011536</td><td>PALB2 Del/Dup</td></tr><tr><td>86011537</td><td>PALB2 Del/Dup Interp</td></tr><tr><td colspan="2">92573-9-Comprehensive Report and Additional Information</td></tr><tr><th>Result Code</th><th>Result Name</th></tr><tr><td>86011542</td><td>Comprehensive Interp</td></tr><tr><td>86011543</td><td>Additional Information</td></tr></table> ***TR (True Reflex) Flag*** ***CPU interface clients: If you are set up to use our True Reflexing option, build the unit codes with the TR flag (indicated above) separately.***
Additional Information	**If the Interpretation Summary is not one of the following three results:** **Positive for a known Pathogenic Mutation;** **Positive for a likely Pathogenic Mutation;** **Positive for a likely Pathogenic Mutation and a variant with unknown significance,** **then TP53, PTEN, CDH1, STK11, and PALB2 will be performed at an additional charge (add CPT code(s) 81321, 81323, 81404, 81405 (x2), 81406 (x2), 81479).** Pricing Message: Negotiated pricing on 91863 will be applied to code 92573 (screen).

Breast Plus Panel without BRCA *(TP53, PTEN, CDH1, STK11, PALB2)*	
Revision Message!	**Please note: The message was updated to remove the New York Patient Testing restriction effective 10/29/14.**
Clinical Significance	**CDH1, PALB2, PTEN, STK11 and TP53 are tumor suppressor genes, where inherited mutations significantly increase breast cancer risk. Testing for mutations in these gene by sequencing and deletion/duplication analysis complement BRCA testing in further elucidate an individual's hereditary breast cancer risk.**
Effective Date	**11/3/2014**
Test Code	**92586**
CPT Codes	**81321, 81323, 81404, 81405 (x2), 81406 (x2), 81479**
Specimen Requirements	**4 mL (2 mL minimum) whole blood collected in an EDTA (lavender-top) tube**
Reject Criteria	**Gross hemolysis; clotted specimens**
Instructions	**Whole Blood: Normal phlebotomy procedure; specimen stability is crucial. Store and ship room temperature immediately. Do not freeze. Send report of results for family member with known BRCA mutation.**
Transport Temperature	**Room temperature**

Specimen Stability	**Room temperature and Refrigerated: 8 days** **Frozen: Unacceptable**
Set-up/Analytic Time	**Set up: Tues, Thurs, Sat; Report available: 14 days from completed pre-authorization**
Reference Range	**Accompanies report**
Methodology	**Exon Capture, Next Generation Sequencing, Multiplex Ligation-dependent Probe Amplification**
Performing Site	**Quest Diagnostics Nichols Institute, San Juan Capistrano**
Interface Mapping	

92586-1-Interpretation Summary

Result Code	Type	Result Name
86007572	Prompt-Result (no return)	Ethnicity:
86011586		Interpretation Summary

92586-2-TP53 Sequencing and Del/Dup

Result Code	Result Name
86011410	TP53 Sequencing
86011411	TP53 Seq Interp
86011412	TP53 Del/Dup
86011413	TP53 Del/Dup Interp

92586-3-PTEN Sequencing and Del/Dup

Result Code	Result Name
86011520	PTEN Sequencing
86011521	PTEN Seq Interp
86011522	PTEN Del/Dup
86011523	PTEN Del/Dup Interp

92586-4-CDH1 Sequencing and Del/Dup

Result Code	Result Name
86011527	CDH1 Sequencing
86011528	CDH1 Seq Interp
86011529	CDH1 Del/Dup
86011530	CDH1 Del/Dup Interp

92586-5-STK11 Sequencing and Del/Dup

Result Code	Result Name
86011513	STK11 Sequencing
86011514	STK11 Seq Interp
86011515	STK11 Del/Dup
86011516	STK11 Del/Dup Interp

92586-6-PALB2 Sequencing and Del/Dup

Result Code	Result Name

86011534	**PALB2 Sequencing**
86011535	**PALB2 Seq Interp**
86011536	**PALB2 Del/Dup**
86011537	**PALB2 Del/Dup Interp**

92586-7-Comprehensive Report and Additional Information

Result Code	Result Name
86011587	**Comprehensive Interp**
86011588	**Additional Information**

CDH1 Sequencing and Deletion/Duplication	
Revision Message!	**Please note: The message was updated to remove the New York Patient Testing restriction effective 10/29/14.**
Clinical Significance	**Germline mutations in the CDH1 (E-cadherin gene) gene have been reported in families with a hereditary predisposition to breast cancer and gastric cancer. Sequencing and deletion/duplication analyses of the CDH1 gene will identify individuals at risk for hereditary breast and gastric cancer.**
Effective Date	**11/3/2014**
Test Code	**92568**
CPT Codes	**81406, 81479**
Specimen Requirements	**4 mL (2 mL minimum) whole blood collected in an EDTA (lavender-top) tube**
Reject Criteria	**Gross hemolysis; clotted specimens**
Instructions	**Whole Blood: Normal phlebotomy procedure; specimen stability is crucial. Store and ship room temperature immediately. Do not freeze. Send report of results for family member with known BRCA mutation.**
Transport Temperature	**Room temperature**
Specimen Stability	**Room temperature and Refrigerated: 8 days** **Frozen: Unacceptable**
Set-up/Analytic Time	**Set up: Tues, Thurs, Sat; Report available: 14 days from completed pre-authorization**
Reference Range	**Accompanies report**
Methodology	**Exon Capture, Next Generation Sequencing, Multiplex Ligation-dependent Probe Amplification**
Performing Site	**Quest Diagnostics Nichols Institute, San Juan Capistrano**

Interface Mapping

Result Code	Type	Result Name
86007572	**Prompt-Result (no return)**	**Ethnicity:**
86011526		**Interpretation Summary**
86011527		**CDH1 Sequencing**
86011528		**CDH1 Seq Interp**
86011529		**CDH1 Del/Dup**
86011530		**CDH1 Del/Dup Interp**
86011531		**Comprehensive Interp**

86011532		**Additional Information**

PALB2 Sequencing and Deletion/Duplication	
Revision Message!	**Please note: The message was updated to remove the New York Patient Testing restriction effective 10/29/14.**
Clinical Significance	**Fanconi anemia (subtype FA-N) has been linked to homyzygous or compound heterozgygous mutations in the PALB2 gene. Germline PALB2 mutations increase the risk of breast and pancreatic cancer. Sequencing and deletion/duplication analyses of the PALB2 gene will identify individuals who are affected or carriers of Fanconi anemia (FA-N), as well as who are at risk for hereditary breast and pancreatic cancer.**
Effective Date	**11/3/2014**
Test Code	**92571**
CPT Codes	**81406, 81479**
Specimen Requirements	**4 mL (2 mL minimum) whole blood collected in an EDTA (lavender-top) tube**
Reject Criteria	**Gross hemolysis; clotted specimens**
Instructions	**Whole Blood: Normal phlebotomy procedure; specimen stability is crucial. Store and ship room temperature immediately. Do not freeze. Send report of results for family member with known BRCA mutation.**
Transport Temperature	**Room temperature**
Specimen Stability	**Room temperature and Refrigerated: 8 days** **Frozen: Unacceptable**
Set-up/Analytic Time	**Set up: Tues, Thurs, Sat; Report available: 14 days from completed pre-authorization**
Reference Range	**Accompanies report**
Methodology	**Exon Capture, Next Generation Sequencing, Multiplex Ligation-dependent Probe Amplification**
Performing Site	**Quest Diagnostics Nichols Institute, San Juan Capistrano**
Interface Mapping	

Result Code	Type	Result Name
86007572	**Prompt-Result (no return)**	**Ethnicity:**
86011533		**Interpretation Summary**
86011534		**PALB2 Sequencing**
86011535		**PALB2 Seq Interp**
86011536		**PALB2 Del/Dup**
86011537		**PALB2 Del/Dup Interp**
86011538		**Comprehensive Interp**
86011539		**Additional Information**

PTEN Sequencing and Deletion/Duplication	
Revision Message!	**Please note: The message was updated to remove the New York Patient Testing restriction effective 10/29/14.**

Clinical Significance	**Defects in the PTEN gene are the main cause of Cowden disease which is characterized by hamartomatous polyps of the gastrointestinal tract, mucocutaneous lesions, and increased risk of developing neoplasms. Mutations in the PTEN gene is also associated with a lifetime breast cancer risk of 50%. Sequencing and deletion/duplication analyses of the PALB2 gene will identify individuals who are affected with Cowden disease as well as who are at risk for hereditary cancers.**
Effective Date	**11/3/2014**
Test Code	**92566**
CPT Codes	**81321, 81323**
Specimen Requirements	**4 mL (2 mL minimum) whole blood collected in an EDTA (lavender-top) tube**
Reject Criteria	**Gross hemolysis; clotted specimens**
Instructions	**Whole Blood: Normal phlebotomy procedure; specimen stability is crucial. Store and ship room temperature immediately. Do not freeze. Send report of results for family member with known BRCA mutation.**
Transport Temperature	**Room temperature**
Specimen Stability	**Room temperature and Refrigerated: 8 days** **Frozen: Unacceptable**
Set-up/Analytic Time	**Set up: Tues, Thurs, Sat; Report available: 14 days from completed pre-authorization**
Reference Range	**Accompanies report**
Methodology	**Exon Capture, Next Generation Sequencing, Multiplex Ligation-dependent Probe Amplification**
Performing Site	**Quest Diagnostics Nichols Institute, San Juan Capistrano**
Interface Mapping	

Result Code	Type	Result Name
86007572	Prompt-Result (no return)	Ethnicity:
86011519		Interpretation Summary
86011520		PTEN Sequencing
86011521		PTEN Seq Interp
86011522		PTEN Del/Dup
86011523		PTEN Del/Dup Interp
86011524		Comprehensive Interp
86011525		Additional Information

STK11 Sequencing and Deletion/Duplication	
Revision Message!	**Please note: The message was updated to remove the New York Patient Testing restriction effective 10/29/14.**
Clinical Significance	**Mutations in the STK11 (aka LKB1) gene causes Peutz-Jeghers syndrome (PJS) which is an autosomal dominant disorder with an increased risk of various neoplasms (especially gastrointestinal and breast cancers). Sequencing and deletion/duplication analyses of the STK11 gene will identify individuals who are affected with PJS as well as who are at risk for hereditary gastrointestinal and breast cancers.**
Effective Date	**11/3/2014**
Test Code	**92565**
CPT Codes	**81405, 81404**

Specimen Requirements	**4 mL (2 mL minimum) whole blood collected in an EDTA (lavender-top) tube**
Reject Criteria	**Gross hemolysis; clotted specimens**
Instructions	**Whole Blood: Normal phlebotomy procedure; specimen stability is crucial. Store and ship room temperature immediately. Do not freeze. Send report of results for family member with known BRCA mutation.**
Transport Temperature	**Room temperature**
Specimen Stability	**Room temperature and Refrigerated: 8 days** **Frozen: Unacceptable**
Set-up/Analytic Time	**Set up: Tues, Thurs, Sat; Report available: 14 days from completed pre-authorization**
Reference Range	**Accompanies report**
Methodology	**Exon Capture, Next Generation Sequencing, Multiplex Ligation-dependent Probe Amplification**
Performing Site	**Quest Diagnostics Nichols Institute, San Juan Capistrano**
Interface Mapping	

Result Code	Type	Result Name
86007572	Prompt-Result (no return)	Ethnicity:
86011512		Interpretation Summary
86011513		STK11 Sequencing
86011514		STK11 Seq Interp
86011515		STK11 Del/Dup
86011516		STK11 Del/Dup Interp
86011517		Comprehensive Interp
86011518		Additional Information

TP53 Sequencing and Deletion/Duplication	
Revision Message!	**Please note: The message was updated to remove the New York Patient Testing restriction effective 10/29/14.**
Clinical Significance	**Mutations in the TP53 gene leads to Li-Fraumeni syndrome (LFS) which is an inherited cancer syndrome with an early onset of tumors, multiple tumors within an individual, and multiple affected family members. The most common types of tumors are soft tissue sarcomas and osteosarcomas, breast cancer, brain tumors, leukemia, and adrenocortical carcinoma. Sequencing and deletion/duplication analyses of the TP53 gene will identify individuals who are affected with LFS and hereditary cancers.**
Effective Date	**11/3/2014**
Test Code	**92560**
CPT Codes	**81405, 81479**
Specimen Requirements	**4 mL (2 mL minimum) whole blood collected in an EDTA (lavender-top) tube**
Reject Criteria	**Gross hemolysis; clotted specimens**
Instructions	**Whole Blood: Normal phlebotomy procedure; specimen stability is crucial. Store and ship room temperature immediately. Do not freeze. Send report of results for family member with known BRCA mutation.**
Transport Temperature	**Room temperature**
Specimen Stability	**Room temperature and Refrigerated: 8 days**

	Frozen: Unacceptable
Set-up/Analytic Time	**Set up: Tues, Thurs, Sat; Report available: 14 days from completed pre-authorization**
Reference Range	**Accompanies report**
Methodology	**Exon Capture, Next Generation Sequencing, Multiplex Ligation-dependent Probe Amplification**
Performing Site	**Quest Diagnostics Nichols Institute, San Juan Capistrano**
Interface Mapping	

Result Code	Type	Result Name
86007572	**Prompt-Result (no return)**	**Ethnicity:**
86011409		**Interpretation Summary**
86011410		**TP53 Sequencing**
86011411		**TP53 Seq Interp**
86011412		**TP53 Del/Dup**
86011413		**TP53 Del/Dup Interp**
86011414		**Comprehensive Interp**
86011415		**Additional Information**