

**TRIPEPTIDYL PEPTIDASE 1 (TPP1) AND PAMITOYL-PROTEIN  
THIOESTERASE (PPT1), FIBROBLASTS  
#89540**

**USEFUL FOR:** Evaluation of patients with clinical presentations suggestive of Neuronal Ceroid Lipofuscinoses (NCL).  
An aid in the differential diagnosis of infantile and late infantile NCL.

**METHODOLOGY:** Fluorometric Enzyme Assay

**REFERENCE VALUES:** TPP1: 69-934 nmol/hr/mg protein  
PPT1: 30-194 nmol/hr/mg protein

**SPECIMEN REQUIREMENTS:** Submit only one of the following specimens:

**Cultured Fibroblasts**

1 T-75 or 2 T-25 flasks filled to neck with culture media.  
Maintain sterility and forward promptly at ambient temperature.  
**(Specimen received in formalin or fixative preservative is not acceptable.)**

**Skin Biopsy**

Collect sterile skin biopsy (4 mm punch) in a sterile, screw-capped container filled with any standard cell culture media (ex. Minimal essential media, RPMI 1640, etc.). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin [Supply T-115]). Maintain stability and forward promptly.  
**(Specimen received in formalin or fixative preservative is not acceptable. Specimen cannot be frozen.)**

**NOTE:** When an order is filed for this test, #8482 Fibroblast Culture and #88832 Cryopreservation for Biochemical Studies are automatically added by the system.

**CAUTIONS:** This assay does not detect carrier status of NCL.

**LIST FEE:** \$ 271.30                      **CPT CODE:** 82657

**ANALYTIC TIME:** 30-45 days                      **DAY(S) SET-UP:** Varies

**QUESTIONS:** Contact your Mayo Medical Laboratories' Regional Manager or  
Sara Siewert, Mayo Medical Laboratories' Technologist Support  
Telephone: 800-533-1710

## TEST DEFINITION

8/21/2009

Code Name  
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89540 TPP1 and PPT1, Fibroblasts

\*\*\*\* End of Name to order code Report \*\*\*\*

MML MML Test setup information

ORDER CODE	EFF DATE	TC	TITLE	Checking Normals	Print normals (# coded)	Perform Site *
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89540	7/2/2009		TPP1 and PPT1, Fibroblasts			MCR
			Transport temp : Varies			
			Ambient\Refrig <24 hours OK\Frozen NO-Cultured Fibroblasts			
			Refrig\Ambient OK\Frozen NO-Skin Biopsy			
		50791	Specimen			
		50792	Specimen ID			
		50793	Source			
		50794	Order Date			
		50795	Reason for Referral			
		50796	Method			
		50797	TPP1, F			
			Units: nmol/hr/mg prot			
			NO SEX			
			All Ages : 69-934		; 69-934	
			MALE			
			All Ages : 69-934		; 69-934	
			FEMALE			
			All Ages : 69-934		; 69-934	

(continued next page)

MML MML Test setup information

ORDER CODE -----	EFF DATE -----	TC ----	TITLE -----	Checking Normals -----	Print normals (# coded) -----	Perform Site * -----
89540	(continued)					
		50798	PPT1, F	Units: nmol/hr/mg prot		
			NO SEX			
			All Ages : 30-194		; 30-194	
			MALE			
			All Ages : 30-194		; 30-194	
			FEMALE			
			All Ages : 30-194		; 30-194	
		50799	Interpretation			
		50800	Amendment			
		50801	Reviewed By			
		50802	Release Date			

\*Performing Site Legend

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MCR      Mayo Clinic Dpt of Lab Med & Pathology      LAB DIRECTOR:  Franklin R. Cockerill, III, M.D.
          200 First Street SW
          Rochester, MN 55905
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\*\*\* End of Report \*\*\*

MML Messages used as normals

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CODE      TEXT
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Total of 0 normals codes

\*\*\* End of Report \*\*\*



# TEST DEFINITION

6/15/2009

MML NAME/ORDER CODE CROSS-REFERENCE

MML CODE	NAME
8482	FIBROBLAST CULTURE

MML TEST SETUP INFORMATION

ORDER CODE	EFF DATE	TC	TITLE	CHECKING NORMALS	PRINT NORMALS (# CODED)	PERFORM SITE *
8482	1/31/2002		FIBROBLAST CULTURE			MCR

TRANSPORT TEMP : REFRIG\AMBIENT OK\FROZEN NO  
 8482 FIBROBLAST CULTURE

POSSIBLE RESULT VALUES INCLUDE : CELLS IN CULTURE, RESULTS TO FOLLOW.  
 NO SEX  
 ALL AGES : ; NOT APPLICABLE  
 MALE  
 ALL AGES : ; NOT APPLICABLE  
 FEMALE  
 ALL AGES : ; NOT APPLICABLE

\*PERFORMING SITE LEGEND

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MCR	MAYO CLINIC DPT OF LAB MED & PATHOLOGY	LAB DIRECTOR:	FRANKLIN R. COCKERILL, III, M.D.
	200 FIRST STREET SW		
	ROCHESTER, MN 55905		

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MML MESSAGES USED AS NORMALS

MML CODE	TEXT
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TOTAL OF 0 NORMALS CODES

\*\*\* END OF REPORT \*\*\*



# TEST DEFINITION

11/24/2008

MML Name/Order Code Cross-reference  
Code Name  
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88832 Cryopreserve for Biochem Studies

MML MML Test Setup Information

ORDER CODE	EFF DATE	TC	TITLE	Checking Normals	Print normals (# coded)	Perform Site *
88832	4/16/2007		Cryopreserve for Biochem Studies			MCR
		88832	Cryopreserve for Biochem Studies			

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\*Performing Site Legend

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MCR	Mayo Clinic Dpt of Lab Med & Pathology	LAB DIRECTOR:	Franklin R. Cockerill, III, M.D.
	200 First Street SW		
	Rochester, MN 55905		

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MML Messages used as normals  
CODE TEXT  
-----

Total of 0 normals codes

\*\*\* End of Report \*\*\*

1-800-533-1710

<b>PATIENT NAME</b> TESTING, 89540		<b>PATIENT NUMBER</b>		<b>AGE</b> 1	<b>SEX</b> F	<b>ACCESSION #</b> G9133125
<b>ORDERING PHYSICIAN</b>		<b>CLIENT ORDER #</b>			<b>ACCOUNT #</b> LIAISONS	
<b>COLLECTION</b> 08/06/09 12:44 P <b>DATE TIME</b>	<b>RECEIVED</b> 08/06/09 12:44 P <b>DATE TIME</b>	<b>REPORT PRINTED</b> 08/21/09 08:57 A <b>DATE TIME</b>		<b>SPECIMEN INFORMATION</b> DATE OF BIRTH:		
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
<b>Fibroblast Culture</b>			
Fibroblast Culture	Testing	Not applicable	MCR
<b>Cryopreserve for Biochem Studies</b>			
Cryopreserve for Biochem Studies	Testing		MCR
<b>TPP1 and PPT1, Fibroblasts</b>			
Specimen	Fibroblasts		MCR
Specimen ID	1038465		MCR
Order Date	06 Aug 2009 12:57		MCR
Reason for Referral			MCR
Reason for referral not provided.			
Method			MCR
Fluorometric Enzyme Assay			
TPP1, F	98	nmol/hr/mg prot	69-934 MCR
PPT1, F	40	nmol/hr/mg prot	30-194 MCR
Interpretation			MCR
<p>In this sample the activity of both Palmitoyl-Protein Thioesterase 1 and Tripeptidyl Thioesterase 1 enzymes are normal. These results indicate that this individual does not have a defect in either the CLN1 or CLN2 gene. However, this does not rule out other forms of neuronal ceroid lipofuscinosis. If clinically indicated, additional testing by Electron Microscopy (MML # 4993) is recommended. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) to determine the best testing strategy or if you have any questions.</p>			
Reviewed By	Kimiyo M Raymond MD		MCR
Release Date	06 Aug 2009 14:52		MCR

\* Perform Site Legend on last page of report

<b>PATIENT NAME</b> TESTING, 89540	<b>ORDER STATUS</b> Final	<b>COLLECTION DATE AND TIME</b> 08/06/09 12:44 P
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1-800-533-1710

<b>PATIENT NAME</b> TESTING, 89540		<b>PATIENT NUMBER</b>		<b>AGE</b> 1	<b>SEX</b> F	<b>ACCESSION #</b> G9133125
<b>ORDERING PHYSICIAN</b>		<b>CLIENT ORDER #</b>				<b>ACCOUNT #</b> LIAISONS
<b>COLLECTION</b> 08/06/09 12:44 P	<b>RECEIVED</b> 08/06/09 12:44 P	<b>REPORT PRINTED</b> 08/21/09 08:57 A		<b>SPECIMEN INFORMATION</b> DATE OF BIRTH:		
<b>DATE</b> <b>TIME</b>	<b>DATE</b> <b>TIME</b>	<b>DATE</b> <b>TIME</b>				
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

<b>TEST REQUESTED</b>	<b>HI</b> <b>LO</b>	<b>REF RANGE</b>	<b>PERFORM SITE *</b>
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## \* PERFORMING SITE

MCR      Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW    Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.
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<b>PATIENT NAME</b> TESTING, 89540	<b>ORDER STATUS</b> Final	<b>COLLECTION DATE AND TIME</b> 08/06/09 12:44 P
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