



PATIENT'S INFORMATION

Last Name	First Name	M.I.	D.O.B.(mm/dd/yyyy) / /	SEX <input type="checkbox"/> M <input type="checkbox"/> F	DIAGNOSIS: ICD-10
Address	Street	Apt. #	Telephone #	FACILITY CONSECUTIVE # / PATIENT ID #	
City	State	Zip			

PERSON RESPONSIBLE FOR BILL (OUTPATIENTS ONLY)

Last Name	First Name	<input type="checkbox"/> INSURANCE CARRIER _____	
		<input type="checkbox"/> INSURANCE I.D. # _____	
		<input type="checkbox"/> MEDICAID CARD REQUIRED	
Address	Street	Apt. #	Telephone #
City	State	Zip	

PHYSICIAN INFORMATION

Last Name	First Name	State	NPI Number
			Facility
Address	Street	Apt. #	City
			State
			Zip
Fax ()	PHYSICIAN'S SIGNATURE		DATE: / /
Telephone ()			

By law, test cannot be performed without physician and patient authorization

Collection Date / /	<input type="checkbox"/> Initial Study <input type="checkbox"/> Follow-up	Specimens Accepted Monday – Friday
------------------------	--	---

<p align="center"><u>PLEASE CHECK TEST(S)</u></p> <p><input type="checkbox"/> Kufs common mutations</p> <p><input type="checkbox"/> Family mutations</p>	<p>SPECIMEN INFORMATION</p> <p><input type="checkbox"/> Whole Blood: One Purple-Top Tube (sodium EDTA)</p> <p><input type="checkbox"/> Submit specimens immediately in sterile tubes on blue ice (not frozen)</p>
---	--

...For Lab Use Only...

Accession Number	Date:	Reviewed By:
Date Rec'd in Lab		
M#		

IBR Specialty Clinical Laboratories

1050 Forest Hill Road, Staten Island, New York 10314-6399

Tel (718) 494-5219 Fax (718) 494-1026

Joseph J. Maturi, M.S.

Acting Director, Institute for Basic Research

Milen Velinov, M.D., Ph.D., DABMG

Director, Specialty Clinical Laboratories

Consent form for genetic testing: Autosomal Dominant Kufs Disease-common mutations

Description of the disease

Autosomal-dominant Kufs disease (ADKD) is a rare disorder that affects the central nervous system. The manifestations usually start after 20 years of age and include memory loss, seizures, behavioral abnormalities, difficulties to keep balance and dementia. Sometimes it is passed from a parent to a child. Some other times it happens as a new gene change in one person in the family.

Description of the test

This test studies the gene called DNAJC5 for the presence of two mutations that were to-date associated with ADKD. It studies only particular region of this gene where the mutations are located.

Principle of the test

The test consists in first making many copies of the studied portion of the gene, and then looking for two particular changes in the DNA sequence in this gene region.

Meaning of a positive test

Positive test result means that one of the investigated gene mutations is present in the tested individual. That means that this individual is affected with ADKD.

Meaning of a negative test

Negative test means that the tested individual does not have any of the common mutations in the gene DNAJC5 that are associated with ADKD. However, this negative result does not rule out ADKD in the individual that is tested since other gene mutations that are not included in the assay may be present in this individual.

Test limitations

This test will not identify all genetic changes associate with ADKD. Therefore, as mentioned above, negative test would not rule out the presence of ADKD.

Confidentiality

The result of this testing will be sent only to the care provider, who ordered the test.

I understand the above and give consent for diagnostic testing.

Signature of Subject/or Parent/Guardian

Date

Physician/Genetic Counselor

Date

IBR Specialty Clinical Laboratories

1050 Forest Hill Road, Staten Island, New York 10314-6399

Tel (718) 494-5219 fax (718) 494-1026

Joseph J. Maturi, M.S.

Acting Director, Institute for Basic Research

Milen Velinov, M.D., Ph.D., DABMG

Director, Specialty Clinical Laboratories

Genetic counseling and additional evaluation

Genetic counseling regarding possible results of this testing will be provided prior to conducting the test and will be available after the testing for all participants. Further testing and/or additional physician consults may be warranted for some of the participants.

Statements about specimen retention

We would be interested in keeping your specimen with your permission after completion of this test for potential use in further research on the molecular basis of ADKD. If you do not give us your permission to keep your specimen for further research, it will be destroyed 60 days after it was obtained. Please write your initials to choose one of the options below:

_____ I agree to have my specimen kept for further research after this test is complete.

_____ No, I do not agree to have my specimen kept for further research, please destroy this after the test is done.

Signature of client

Print name

Signature

Date

Signature of the person obtaining the consent

Print name

Signature

Date