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
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Molecular Testing for Kufs Disease

PATIENT'S INFORMATION

Last Name	First Name	M.I.	D.O.B.(mm/dd/yyyy) / /	SEX ☐M ☐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	☐ INSURANCE CARRIER _____ ☐ INSURANCE I.D. # _____ ☐ 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 ( ) Telephone ( )	PHYSICIAN'S SIGNATURE	DATE: / /	

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

Collection Date / /	☐ Initial Study ☐ Follow-up	**Specimens Accepted Monday – Friday**

PLEASE CHECK TEST(S)	**SPECIMEN INFORMATION**
☐ Kufs common mutations ☐ Family mutations	☐ Whole Blood: One Purple-Top Tube (sodium EDTA) ☐ Submit specimens immediately in sterile tubes on blue ice (not frozen)

…For Lab Use Only…

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

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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

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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 ion 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