

DEPARTMENT OF ENDOCRINOLOGY, DIABETES & METABOLISM CHRISTIAN MEDICAL COLLEGE – VELLORE



Molecular Genetic Diagnosis of Lipodystrophy Syndromes

AGPAT2, BSCL2, INSR, LMNA, PPAR γ & ZMPEST24 PANEL
NEXT GENERATION SEQUENCING BASED STRATEGY

Patient Information

Name:		Hospital No. :	
Address:		Age:	Sex:
		Ethnic Origin:	
		Education:	
Phone:	E-mail:	Occupation:	

History

DM: Yes/No	Initial Type:	Age at diagnosis:	Current treatment: OAD/Insulin
Treatment details:			
Signs of weight loss at onset:		Age at onset:	
Weight gain over abdomen: Yes/No	Progression of weight loss over _____ years		Selective weight loss in limbs: Yes/No
H/O PCOS: Yes/No	Menstrual History: M ___ ___-___d _____d regular/irregular average/increased/decreased flow		
H/O Jaundice: Yes/No	CVS: Normal/Breathlessness/Angina/Syncope/Palpitations/Other		
H/O Hypertension: Yes/No	H/O Frothy urine: Yes/No	H/O Pedal Oedema: Yes/No	
Other relevant history:			
Mentally challenged: No/Mild/Moderate/Severe			
Family history of similar illness: Yes/No	If Yes mention relation:		
Approximate calorie intake per day:	Diet: Veg/Non-veg	Appetite: Normal/Decreased/Increased	
Alcohol consumption: Yes/No	Duration:	Amount per week:	
Smoking: Yes/No	Duration:	Pack years:	

Examination

Initial Weight:	Current weight:	Current Height:	Current BMI:
Pulse: /min	BP: / mm Hg	Icterus: Yes/No	Pedal Oedema: Yes/No
General Appearance: Normal/Cushingoid/Acromegalic/Progeroid/Other		Umbilical prominence: Yes/No	
Muscular hypertrophy: Yes/No	Phlebomegaly: Yes/No	Hirsutism/Virilism: Yes/No	
Acanthosis nigricans: Yes/No	Grade: I/II/III	Hepatomegaly: Yes/No	Abdominal circumference: ___cm
Normal skin: Yes/No If not, elaborate:			
Other findings pertaining to the clinical diagnosis:			

Biochemistry

Haemoglobin:	GRBS:	a. c./p. c.:	HbA1C:	Insulin(fasting):
Insulin(90 min p. c.):	C-peptide(fasting):		C-peptide(90 min p. c.):	LDL:
HDL:	Triglycerides:	Total Cholesterol:	Albumin:	Total Bilirubin:
Direct Bilirubin:	SGOT:	SGPT:	Creatinine:	Urine ACR:

Consent

- I have understood that my blood sample will be used for diagnostic and research purposes relevant to my health condition. Please circle: Yes/No
- I also give my consent to contact me at the address mentioned above regarding my genetic test report and related research. Please circle: Yes/No

Patient's Signature:

Witness' Signature:

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Pedigree

(Draw a pedigree chart with details of 3-4 generations, age of onset, treatment details, complications, proband name, contact no. of affected members, sample code and hospital no.)

Overview

Diabetes Mellitus	Early onset/Late onset/No
Features of insulin resistance	Yes/No
Selective adipose tissue loss	Yes/No
Localised pattern of fat loss	Yes/No
Associated autoimmunity	Yes/No
Pattern of inheritance	Autosomal dominant/autosomal recessive/indefinite/none

Justify if not fitting into the clinical criteria or provide relevant clinical information

Doctor's signature:

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Specimen Shipment Details

Delivery accepted Monday to Friday; patient information sheet should accompany all specimens.

MODY genetic testing requisition is accepted only from a qualified healthcare provider.

Specimen Shipment Details

Collect 5 ml of whole blood in purple top EDTA tube. For small babies, we require a minimum of 1 ml of blood.

Ship blood tubes at 4 degrees temperature (frozen ice pack) in an insulated container. Do not freeze blood.

Label the tube with the patient name, date of birth and/or ID number and the center from which the sample is being sent.

Genomic DNA

Send in a sealed tube with at least 20 μ g of purified DNA at a concentration of at least 100 ng/ μ l.

DNA sample can be shipped at room temperature. Include the details of the 260/280 ratio which should be around 1.8.

Label the tube with patient's name, date of birth, ID number and name of the referral center.

Shipping Address

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For further queries or requests

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References

1. Asha HS, Chapla A, Shetty S, Thomas N (2015) Next-generation sequencing-based genetic testing for familial partial lipodystrophy. AACE Clin Case Rep 1(1):e28–e31
2. Shetty S, Chapla A, Kapoor N, Thomas N, Paul TV. A novel variant of the AGPAT2 mutation in generalized congenital lipodystrophy, detected by Next Generation Sequencing. AMJ 2016;9(6):164–168.
3. Garg A. Acquired and inherited lipodystrophies. N Engl J Med. 2004;350:1220-1234.
4. Brown RJ, Araujo-Vilar D, Cheung PT, Dunger D, Garg A et al., The diagnosis and management of lipodystrophy syndromes: a multi-society practice guideline. J Clin Endo. Metab. 2016;101(12):4500–4511.
5. RIG Holt, C Cockram, A Flyvbjerg, BJ Goldstein. Textbook of Diabetes. Oxford, U.K., Wiley-Blackwell, 2017