DIAGNOSING GAUCHER DISEASE

GAUCHER DISEASE CAN BE DIAGNOSED OR RULED OUT WITH A SIMPLE BLOOD-BASED ASSAY

Does your patient exhibit two or more of the following symptoms?[1,2]

- Splenomegaly
- Thrombocytopenia
- Bone abnormalities
- Anaemia
- Hepatomegaly
- Abnormal liver function

IF YES

STEP 1:

Rule out thalassaemia and malignancies

STEP 2:

PERFORM GAUCHER ENZYME TEST*

Perform the Gaucher enzyme test with dried blood spot (DBS)*[1,2]

The siblings of a diagnosed Gaucher disease patient should also be tested.

THE DRIED BLOOD SPOT ASSAY TESTS FOR LOW LEVELS OF LYSOSOMAL GLUCOCEREBROSIDASE

Glucocerebrosidase levels in Gaucher disease patients are typically at **only 10–15%** of normal levels.[1]

Following a positive enzyme test, diagnosis of Gaucher disease is confirmed by DNA sequencing of the glucocerebrosidase gene.[1]

*Acid ß-glucosidase enzyme activity assay.
DBS: dried blood spot.

For healthcare professionals only

References

1. Mehta A, Kuter DJ, Salek SS, *et al.* Presenting signs and patient co-variables in Gaucher disease: outcome of the Gaucher Earlier Diagnosis Consensus (GED-C) Delphi initiative. *Intern Med J.* 2019;49(5):578-91.
2. Mistry PK, Cappellini MD, Lukina E, *et al.* Consensus conference: A reappraisal of Gaucher disease-diagnosis and disease management algorithms. *Am J Hematol.* 2011;86(1):110.

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sanofi-aventis (Malaysia) Sdn Bhd (334110-P), Unit TB-18-1, Level 18, Tower B, Plaza 33, No. 1 Jalan Kemajuan, Seksyen 13, 46200 Petaling Jaya, Selangor Darul Ehsan, Malaysia.
Tel: 03 7651 0800, Fax: 03 7651 0805.

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