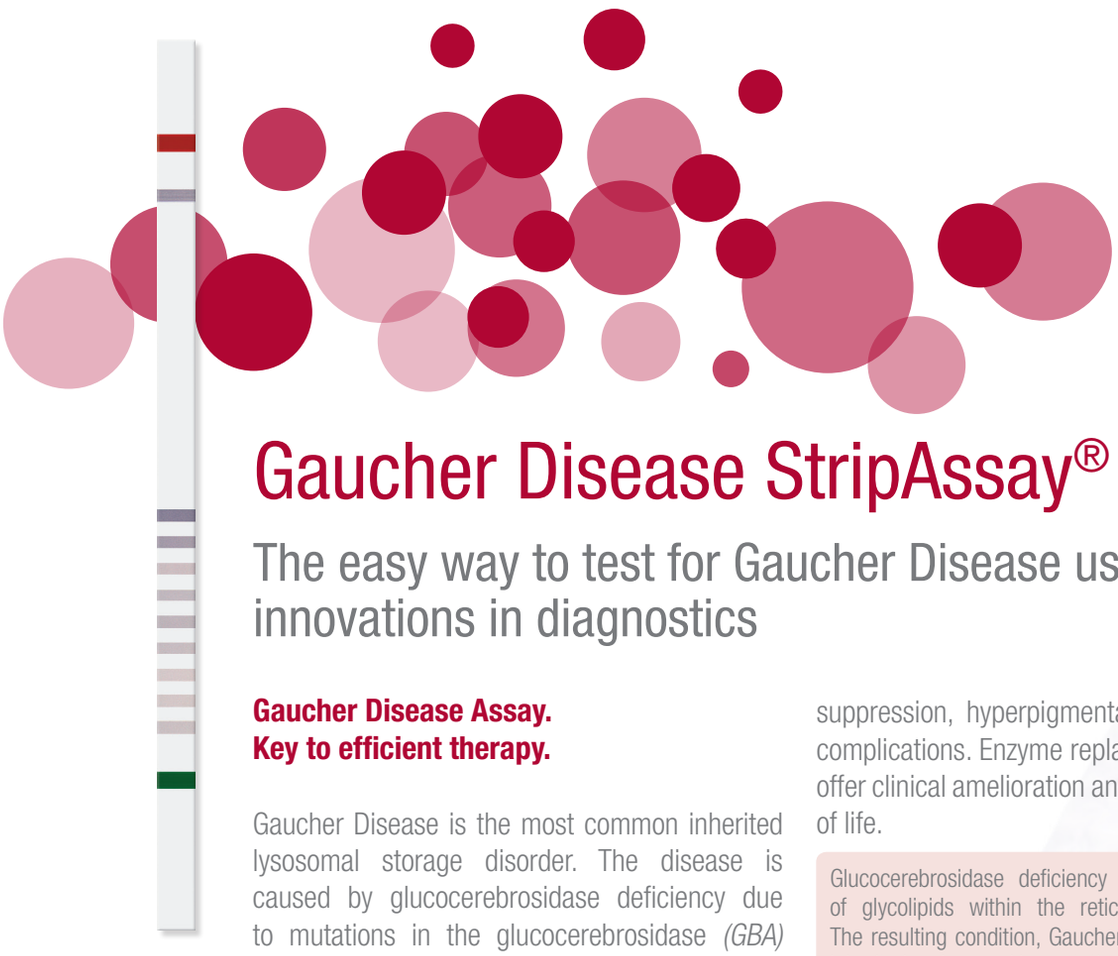
VIENNA LAB
ESTABLISHED INNOVATIONS IN DIAGNOSTICS



Gaucher Disease StripAssay®

The easy way to test for Gaucher Disease using established innovations in diagnostics

Gaucher Disease Assay.
Key to efficient therapy.

Gaucher Disease is the most common inherited lysosomal storage disorder. The disease is caused by glucocerebrosidase deficiency due to mutations in the glucocerebrosidase *(GBA)* gene. Impaired enzyme activity results in a heterogeneous range of clinical manifestations, including enlarged spleen and liver, anemia, thrombocytopenia, bone lesions, bone marrow suppression, hyperpigmentation and neurologic complications. Enzyme replacement therapy may offer clinical amelioration and an improved quality of life.

Glucocerebrosidase deficiency leads to accumulation of glycolipids within the reticulo-endothelial system. The resulting condition, Gaucher Disease, is recessively inherited and caused by mutations in the *GBA* gene. Most of these are point mutations, but some common ones are due to genetic rearrangement between the *GBA* gene and the highly homologous pseudogene.

ViennaLab´s Gaucher Disease StripAssay® offers an easy way to identify the most frequent mutations and recombinant alleles in the *GBA* gene.

Gene	Cellular Function	Status	Therapy	Quality of Life
GBA	Glycolipid metabolism	wildtype		+++
		mutated	✓	++

The Assay

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The ViennaLab Gaucher Disease StripAssay® meets customer requirements

Requirement	ViennaLab's offer
Easy	Three simple steps. 6 h. Done.
Reliable	Probes for variants and controls combined on one teststrip.
Versatile	Automated or manual processing.
Affordable	Incubator.Thermocycler. Shaker. That is all you need. Software for interpretation of results is optional.

ViennaLab´s Gaucher Disease StripAssay®

- is based on reverse-hybridization of biotinylated PCR products
- combines probes for variants and controls in a parallel array of allele-specific oligonucleotides
- works with immobilized oligos on a teststrip
- generates test results by enzymatic color reaction easily visible to the naked eye

Mutations detected

8 common mutations and
2 recombinant alleles in the *GBA* gene
a) 84GG (c.84_85insG), IVS2+1 (c.115+1G>A), N370S (c.1226A>G), V394L (c.1297G>T), D409H (c.1342G>C), L444P (c.1448T>C), R463C (c.1504C>T), R496H (c.1604G>A)
b) rec NciI, rec TL

The three steps of ViennaLab´s Gaucher Disease StripAssay®

Step	Requirement
1. Amplification: Multiplex PCR-amplification. Simultaneous biotin-labeling	Thermocycler
2. Hybridization: Directly on the StripAssay® teststrips	Incubator
3. Identification: Labelled products detected by streptavidin-alkaline phosphatase	Naked eye or scanner & software

Order Information:
Gaucher Disease StripAssay®: 4-250 (20 tests/kit)

Manufacturer:
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