DIFFERENTIAL DIAGNOSIS OF TYPE 1 GAUCHER DISEASE IN PEDIATRIC PATIENTS: KNOW, SUSPECT, AND TEST EARLY FOR IMPROVED OUTCOMES

Type 1 Gaucher disease (GD1) is a rare genetic lysosomal storage disorder in which the enzyme glucocerebrosidase is deficient.[1-3] This leads to an accumulation of the fatty substance glucocerebroside in the affected cells and the formation of 'Gaucher cells'. Over time, Gaucher cells infiltrate various organs including the spleen, liver, and bone marrow, resulting in progressive organ damage.[2,4]

Early diagnosis is critical for managing and slowing disease progression of GD1, yet patients frequently experience considerable diagnostic delays due to its rarity, heterogeneity, and the overlap of GD1 symptoms with several other diseases.[5]

MAJOR SIGNS & SYMPTOMS OF GD[5]

Gastroenterological
- Splenomegaly (unexplained three-fold spleen enlargement)
- Hepatomegaly (mild or moderate deviation)

Hematological
- Anemia (mild or moderate deviation)
- Thrombocytopenia (mild or moderate deviation)
- Gammopathy

Orthopedic
- Bone pain (or more severe bone signs/symptoms)

General Medicine
- Hyperferritinemia (mild or moderate deviation)

COVARIABLES OF GD[5]

Inheritance
- Jewish ancestry
- Family history of GD

Adapted from Weinreb et al. 2022

CHILDHOOD DISEASES WITH SIGNS AND SYMPTOMS OVERLAPPING WITH GD[5]

- Cancers, particularly hematologic cancers
- Hematologic disorders, e.g. idiopathic thrombocytopenia
- Legg–Calvé–Perthes disease
- Sarcoidosis
- Von Willebrand disease
- Allergies and growing pains
- Metabolic bone diseases (multiple causes, e.g. rickets, vitamin C deficiency, copper deficiency, sickle cell disease, Paget's disease)
- Bacterial osteomyelitis
- Other lysosomal storage diseases, e.g. GM1 gangliosidosis, lysosomal acid lipase deficiency, and Niemann–Pick disease type A and C

Adapted from Weinreb et al. 2022

Please see next page for a recommended diagnostic algorithm for GD1.

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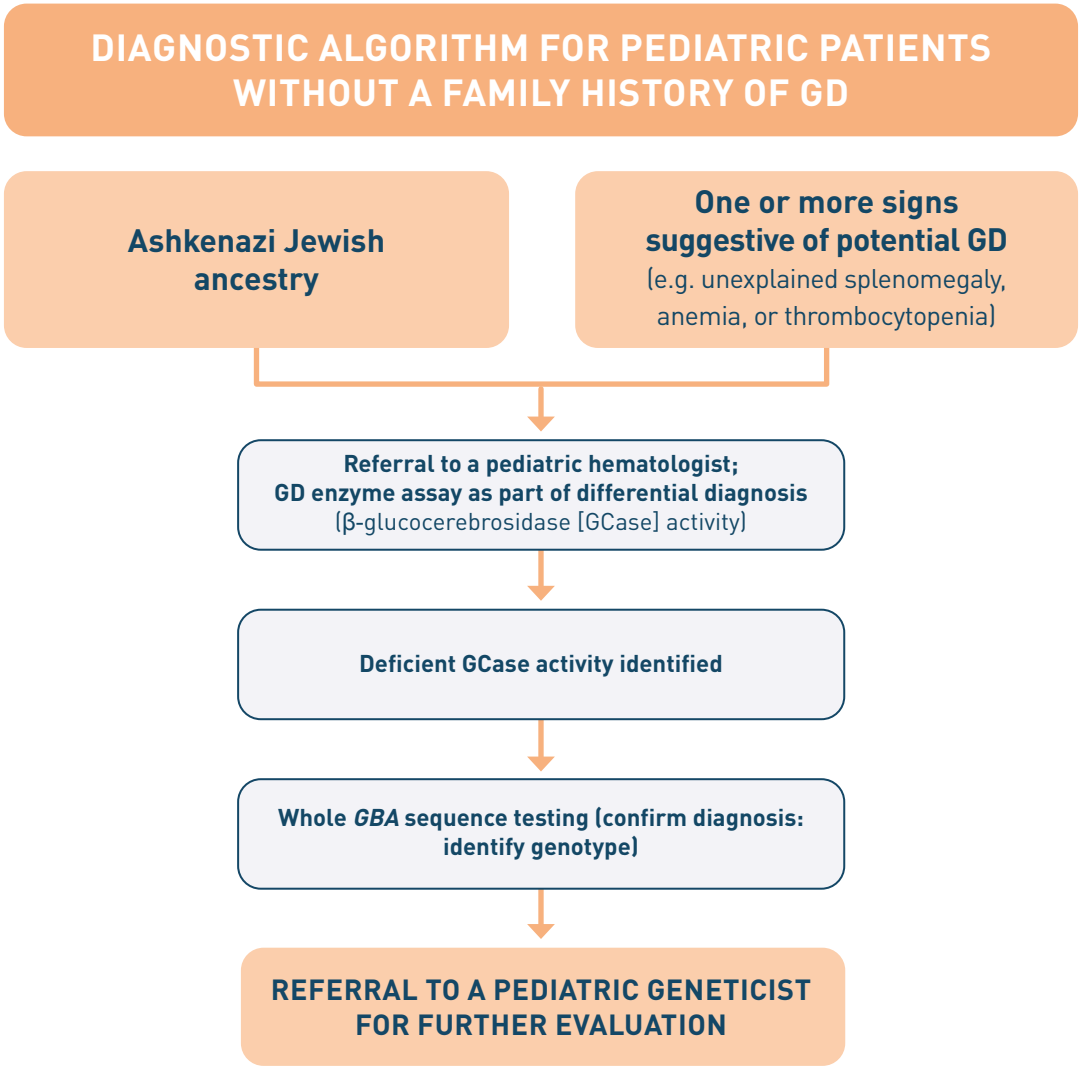
To assist in diagnosis, algorithms have been developed based on recent evidence and current understanding of biomarkers. The following algorithm has been proposed for pediatric patients without a family history of GD, based on a Delphi consensus.[5,6]

This algorithm is not intended to be a diagnostic tool. It does not replace the need for a complete evaluation of the patient by a healthcare professional.



Adapted from Weinreb et al. 2022

To learn more about the signs and symptoms of GD1, and view other useful resources, visit **knowgaucherdisease.com/hcp**

References

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