

Evidence Discussion – Gaucher Disease

(PN 6.3)

EviCore
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- Initial imaging and lifelong re-imaging is supported due to Gaucher Disease progressive, multisystem involvement.^{1, 2, 3}
- Due to bone involvement, including increased risk for Multiple Myeloma, Skeletal X-rays, MRI of Lumbar Spine and MRI Bilateral Femurs are supported.^{1,2,3} Delineating extent of disease can have positive impact on developing appropriate treatment strategies
- Dual-energy x-ray absorptiometry (DXA) Scan is supported to evaluate for bone disease including increased risk of osteoporosis.^{1, 2, 3} This study helps to predict and avoid pathologic fractures.
- CT is the preferred study for the evaluation of lung parenchyma and is supported to evaluate for pulmonary involvement.^{1, 2, 3}
- MRI Abdomen is supported to evaluate for associated visceral disease, such as hepatic, splenic and biliary disease.^{1, 2, 3} This Modality has better signal to noise ratio and soft tissue contrast helping to make more precise diagnosis.
- The role of PET/CT imaging in Gaucher Disease is yet to be established. In the absence of malignancy, PET/CT is not considered medically necessary in the evaluation of Gaucher disease.³ Unnecessary use of this study would expose the patient to excess radiation and noncontributory imaging.

1. Hughes DA, Pastores GM. Gaucher Disease. In: Adam MP, Feldman J, Mirzaa GM, et al., eds. *GeneReviews*®. Seattle (WA): University of Washington, Seattle; July 27, 2000.
2. Degnan AJ, Ho-Fung VM, Ahrens-Nicklas RC, et al. Imaging of non-neuronopathic Gaucher disease: recent advances in quantitative imaging and comprehensive assessment of disease involvement. *Insights Imaging*. 2019;10(1). doi: 10.1186/s13244-019-0743-5.
3. Simpson WL, Hermann G, Balwani M. Imaging of Gaucher Disease. *World J Radiol*. 2014;6(9):657-668. doi: 10.4329/wjr.v6.i9.657