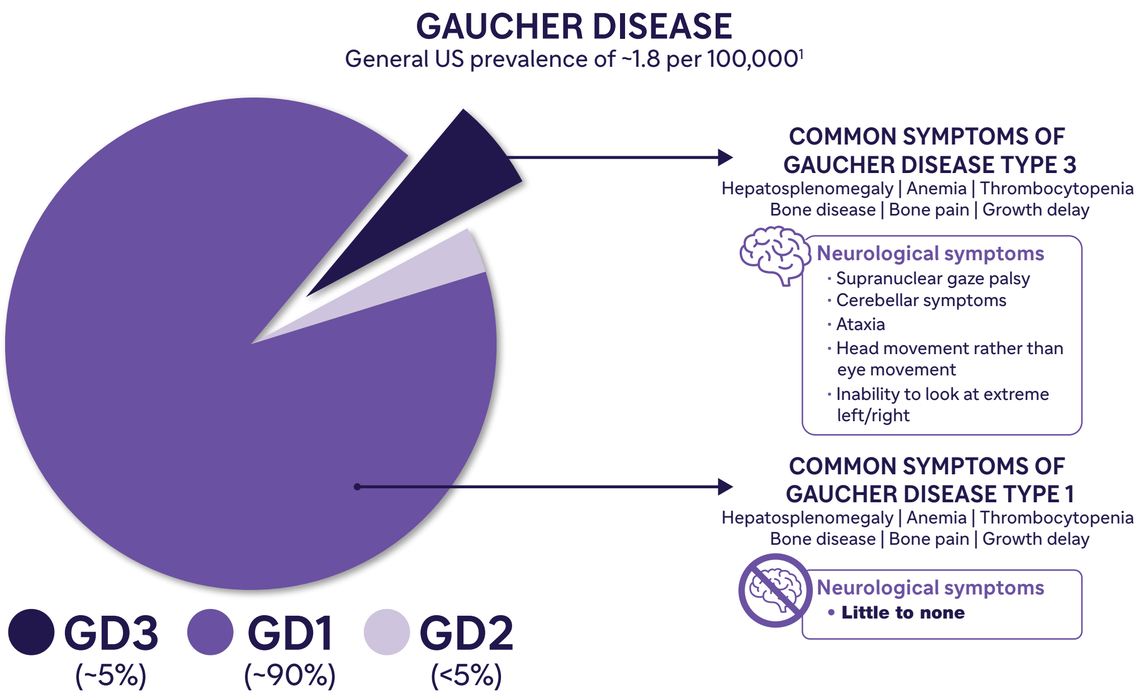
Gaucher Disease Type 3: A Niche Within a Niche

Understanding GD3 and the spectrum of Gaucher disease

Gaucher disease (GD) is a **phenotypic continuum** with a spectrum of clinical presentations.

The clinical phenotype of GD can be classified based on **absence** (type 1) or **presence** (types 2 & 3) of neurological deficits.



Variability in neurologic manifestations may result in the potential misclassification of GD3 as GD1

In a 2009 study from Japan,

>40% of patients with **GD3** were initially misclassified[3]

NOTE: Prevalence percentage of GD3 in Japan is higher than in US.

Disease burden begins early in life[2]

GD3 can present over different life stages and show **aggressive systemic involvement**, with ataxia symptoms more prevalent than neurocognitive ones in patients <5 years of age.

GD3 patient journey

Systemic signs and symptoms + **Neuronal dysfunction beginning in childhood**

Systemic (non-neuronopathic) signs and symptoms >50%, <5 years of age
- Splenomegaly
- Hepatomegaly
- Thrombocytopenia
- Anemia
- Growth Delay

Neuromuscular and neurocognitive signs and symptoms >50%[a]

Newborns/infants (0-1 year)	Toddlers (1-3 years)	Preschoolers (3-5 years)
Cerebellar symptoms (trouble with balance/ wide-based gait)		
Ataxia		
Inability to look extreme left/right	Head movement rather than eye movement	
	Head thrusting	
	Retroflexion of the head/ spinal alignment	
Supranuclear gaze palsy		

GD3 symptomology is diverse and heterogenous—however **all patients** with GD3 show signs of horizontal supranuclear gaze palsy. Signs of ataxia are also common in children ≤5 years of age.

[a]Life stage (by age) when the sign or symptom was first identified or described.

NOTE: A more extensive list of systemic and neuronopathic symptoms occurring <50% of the time—including those that occur later in life—can be found here.

Current management strategies for GD3

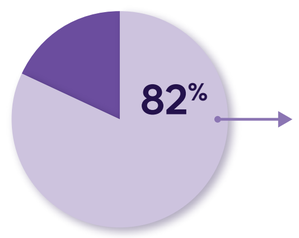
Although there is treatment available for systemic elements of GD3, there are no treatments approved to address neurological manifestations

Investigational agents are being studied in clinical trials to determine how to address neurological symptoms, including those related to ataxia.

Economics of symptom management

Managing symptoms of patients with GD3 can present multiple challenges. **HCRU can be substantial**, including challenges with polypharmacy based on heterogeneity of symptoms.

82% of patients visited at least one medical specialist after diagnosis[4]

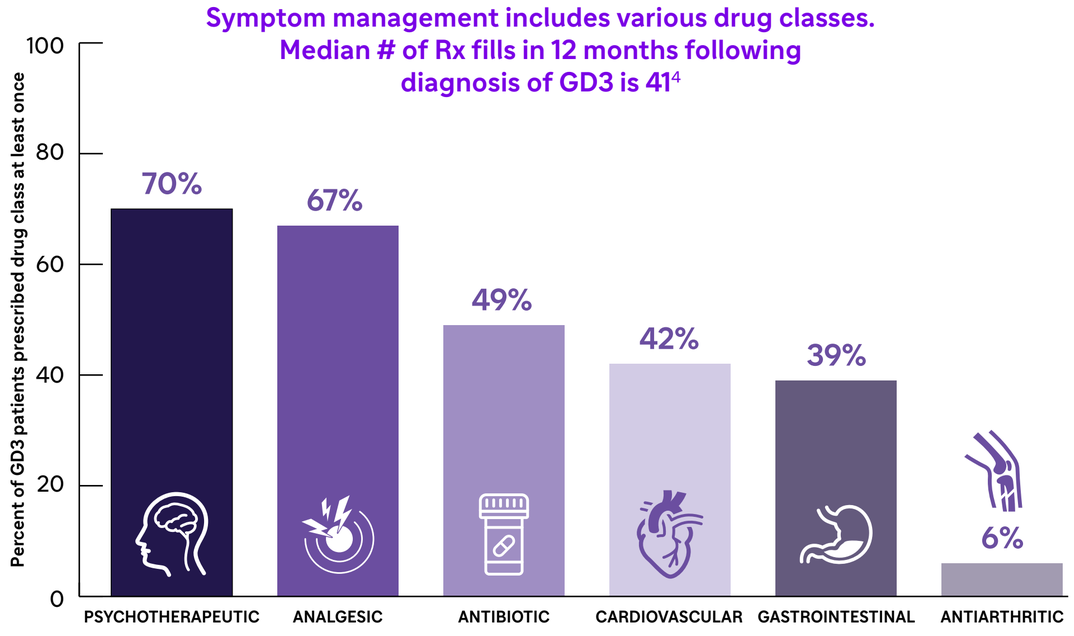


Common specialists seen include neurologists, internists, oncologists, and gastroenterologists

33% of records showed an ER visit within 12 months of diagnosis[4]

Median number of specialist visits over a 3+ year time span: 94[4]

Symptom management includes various drug classes. Median # of Rx fills in 12 months following diagnosis of GD3 is 41[4]



HCRU, healthcare resource utilization.

GD3: A rare disease in need of additional solutions

For additional information, please contact your Sanofi account representative.

References: 1. Rossi C, Ferrante R, Valentinuzzi S, et al. Non-invasive DBS-based approaches to assist clinical diagnosis and treatment monitoring of Gaucher disease. *Biomedicines*. 2023;11(10):2672. **2.** Schiffmann R, Turnbull J, Krupnick R, et al. Gaucher disease type 3 from infancy through adulthood: a conceptual model of signs, symptoms, and impacts associated with ataxia and cognitive impairment. *Orphanet J Rare Dis*. 2025;20:171. **3.** Tajima A, Yokoi T, Ariga M, et al. Clinical and genetic study of Japanese patients with type 3 Gaucher disease. *Mol Genet Metab*. 2009;97:272-277. **4.** Rochmann C, Nanaï A, Bianculli P, et al. Describing the natural history and burden of illness in patients with Gaucher's disease type 3 using a cluster from optum's de-identified Market Clarity Data (2007–2020). Poster presented at: WORLD Symposium, February 25, 2023; Orlando, FL.

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