



Preparing for the Gene Therapy Era: Estimating Rare Disease Prevalence and Eligibility for Gene Therapies in Minnesota, 2018-2022

SUPPLEMENT

JUNE 2026

This Supplement provides details on the process of selecting rare diseases for inclusion in the analysis, the case definition, and the definition of treatment eligibility for the included diseases.

Selection of rare diseases

The Minnesota Department of Health (MDH) started with a list of 16 rare diseases provided by the Minnesota Rare Disease Advisory Council (MNRDAC), which included conditions with at least one of the following: an FDA-approved gene therapy, a gene therapy pending FDA-approval, or a clinical trial for a gene therapy:

- Adrenoleukodystrophy (ALD)
- Batten disease
- Beta Thalassemia
- Delta-Beta Thalassemia
- Duchenne Muscular Dystrophy (DMD)
- Epidermolysis Bullosa
- Hemophilia A
- Hemophilia B
- Hereditary Retinal Dystrophy
- Krabbe Disease
- Metachromatic Leukodystrophy (MLD)
- Rett's Syndrome
- Sickle Cell Disease (SCD)
- Sickle-Beta Thalassemia
- Spinal Muscular Atrophy (SMA)

SUPPLEMENT

PREPARING FOR THE GENE THERAPY ERA: ESTIMATING RARE DISEASE PREVALENCE AND ELIGIBILITY FOR GENE THERAPIES IN MINNESOTA, 2018-2022

MDH evaluated each disease for the following criteria:

- FDA-approved gene therapy or gene therapy in late-stage trials
- Ability to identify cases in claims data

MDH excluded conditions for the following reasons:

- They did not have an FDA-approved gene therapy or in late-stage trials (Batten Disease, Krabbe Disease, Rett's Syndrome).
- There were issues identifying the conditions in claims data (Hereditary Retinal Dystrophy, Delta-Beta Thalassemia, Sickle-Beta Thalassemia).

The following nine conditions met the study conditions and were included in the analysis:

- Adrenoleukodystrophy (ALD)
- Beta Thalassemia
- Duchenne Muscular Dystrophy (DMD)
- Dystrophic Epidermolysis Bullosa (DEB)
- Hemophilia A
- Hemophilia B
- Metachromatic Leukodystrophy (MLD)
- Sickle Cell Disease (SCD)
- Spinal Muscular Atrophy (SMA)

Case definitions and treatment eligibility

All case definitions are based on two ICD-10 diagnosis codes over five years (2018-2022) being present, unless otherwise noted. Two conditions (Beta Thalassemia and Sickle Cell Disease) have different case definitions due to the existence of more specific case definitions based on claims data in the literature.

Case definition and eligibility for treatment with gene therapy

Conditions	Case Definition: Population	Case Definition: Diagnosis and Procedure Codes	Gene Therapy Treatment Eligibility: Include from Cases	Gene Therapy Treatment Eligibility: Exclude if
Adrenoleukodystrophy (ALD)	All	E71.529	Boys, ages 4-17 years ¹	Patients with history of stem cell/bone marrow transplant (CPT: 38240, S2142, S2150; ICD-10: M31.11, T86.0x, T86.5, Z48.290, Z94.81, Z94.84)
Beta Thalassemia	All	One inpatient stay or two outpatient visits with ICD-10 code D56.1 Exclude patients with a code for Alpha Thalassemia [D56.0] or Thalassemia Minor [D56.3] or Other thalassemias [D56.8] or Hereditary persistence of fetal hemoglobin [D56.4] or	Transfusion-dependent beta thalassemia cases (Cases with eight or more red blood cell transfusions [CPT 36430, 36440, 36450, 36455, 36456, 36512, 86999] in the 12 months following beta thalassemia diagnosis), age 4 or older	

SUPPLEMENT

PREPARING FOR THE GENE THERAPY ERA: ESTIMATING RARE DISEASE PREVALENCE AND ELIGIBILITY FOR GENE THERAPIES
IN MINNESOTA, 2018-2022

Conditions	Case Definition: Population	Case Definition: Diagnosis and Procedure Codes	Gene Therapy Treatment Eligibility: Include from Cases	Gene Therapy Treatment Eligibility: Exclude if
		Sickle Cell Disease [D57.0x, D57.1, D572.2x, D57.4x, D57.8x] or Stem cell/bone marrow transplant [CPT: 38240, S2142, S2150; ICD-10: M31.11, T86.0x, T86.5, Z48.290, Z94.81, Z94.84]		
Duchenne Muscular Dystrophy (DMD)	Patients under age 31, with at least two DMD ICD-10 codes before age 18	G71.01	All cases, ages 4-30 years	
Dystrophic Epidermolysis Bullosa (DEB)	All	DEB: Q81.2 but not Q81.1 Epidermolysis Bullosa Other/Unspecified: Q81.8, Q81.9 and no DEB diagnosis code (Q81.2)	All cases (Patients with mutations in the collagen type VII alpha 1 chain (COL7A1) gene are eligible for treatment. However, test results cannot be ascertained from the data.)	
Hemophilia A	All	D66	At least one prescription of emicizumab (CPT J7170,	

SUPPLEMENT

PREPARING FOR THE GENE THERAPY ERA: ESTIMATING RARE DISEASE PREVALENCE AND ELIGIBILITY FOR GENE THERAPIES IN MINNESOTA, 2018-2022

Conditions	Case Definition: Population	Case Definition: Diagnosis and Procedure Codes	Gene Therapy Treatment Eligibility: Include from Cases	Gene Therapy Treatment Eligibility: Exclude if
			Q9995) and/or one prescription for Factor VIII in medical (J7185-J7187, J7190-J7192) or pharmacy claims. Medical claims for Factor VIII must include a D66 code.	
Hemophilia B	All	D67	Cases 18 years old and over with R58 (hemorrhage) OR R23.3 (spontaneous ecchymoses) OR Factor IX (CPT J7193-J7195 or in pharmacy claims)	
Metachromatic Leukodystrophy (MLD)	Patients with MLD ICD-10 before age 8	E75.25	Cases under age 8 years ²	Patients with advanced MLD symptoms (at least one of the following): G-tubes (CPT: 43246, 43762, 43763, 49440, 49452), Prescription for seizure medications,

SUPPLEMENT

PREPARING FOR THE GENE THERAPY ERA: ESTIMATING RARE DISEASE PREVALENCE AND ELIGIBILITY FOR GENE THERAPIES
IN MINNESOTA, 2018-2022

Conditions	Case Definition: Population	Case Definition: Diagnosis and Procedure Codes	Gene Therapy Treatment Eligibility: Include from Cases	Gene Therapy Treatment Eligibility: Exclude if
				Gabapentin prescription, Recipients of stem cell/bone marrow transplant (CPT: 38240, S2142, S2150; ICD-10: M31.11, T86.0x, T86.5, Z48.290, Z94.81, Z94.84)
Sickle Cell Disease (SCD) ³	All	Patients with three SCD diagnosis codes (D57.0, D57.0x, D57.1, D57.2x, D57.4x, D57.8x) within five years	Cases age 12 years and over with a history of pain crises (at least two instances of D57.0x, D57.21)	Recipients of stem cell transplants (ICD-10: M31.11, T86.0x, T86.5, Z48.290, Z94.81, Z94.84; CPT: 38240, S2142, S2150)
Spinal muscular atrophy (SMA)	Patients diagnosed before age 2	G12.0, G12.1, G12.9	Cases under age 2 years	Exclude motor neuron disorders (G12, G12.2, G12.20, G12.24, G12.25, G12.29, G12.8)

SUPPLEMENT

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¹ Skysona (elivaldogene autotemcel) is FDA-approved for early, active cerebral adrenoleukodystrophy (CALD) (source: U.S. Food & Drug Administration. (2022, Sep 16). [BL 125755/0 – Accelerated BLA Approval \(PDF\) \(https://www.fda.gov/media/161665/download?attachment.pdf\)](https://www.fda.gov/media/161665/download?attachment.pdf). Retrieved Dec 15, 2025. CALD cannot be ascertained from claims data, therefore, treatment eligible ALD cases are likely an overestimate.

² Lenmeldy (atidarsagene autotemcel) is FDA-approved for children with pre-symptomatic late infantile, pre-symptomatic early juvenile or early symptomatic early juvenile metachromatic leukodystrophy (source: U.S. Food & Drug Administration. (2024, Mar 18). [FDA Approves First Gene Therapy for Children with Metachromatic Leukodystrophy \(https://www.fda.gov/news-events/press-announcements/fda-approves-first-gene-therapy-children-metachromatic-leukodystrophy\)](https://www.fda.gov/news-events/press-announcements/fda-approves-first-gene-therapy-children-metachromatic-leukodystrophy) [Press release]. This diagnosis involves complex clinical judgment that cannot be reflected in analysis of claims data. Therefore, the number of treatment-eligible cases from the analysis is likely higher than the actual number of patients that would eventually receive gene therapy.

³ Snyder, A. B., Zhou, M., Theodore, R., Quarmyne, M. O., Eckman, J., & Lane, P. A. (2019). [Improving an Administrative Case Definition for Longitudinal Surveillance of Sickle Cell Disease \(https://doi.org/10.1177/0033354919839072\)](https://doi.org/10.1177/0033354919839072). *Public Health Rep*, 134(3), 274-281.

Singh, A., Sontag, M. K., Zhou, M., Dasgupta, M., Crume, T., McLemore, M., Galadanci, N., Randall, E., Steiner, N., Brandow, A. M., Koch, K., Field, J. J., Hassell, K., Snyder, A. B., & Kanter, J. (2023). [Evaluating the Discriminatory Ability of the Sickle Cell Data Collection Program's Administrative Claims Case Definition in Identifying Adults With Sickle Cell Disease: Validation Study \(https://doi.org/10.2196/42816\)](https://doi.org/10.2196/42816). *JMIR Public Health Surveill*, 9, e42816.