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

AN ANALYSIS OF THE MINNESOTA ALL PAYER CLAIMS DATABASE (MN APCD)

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Key findings

- A substantial number of Minnesotans with rare diseases may benefit from gene therapy. This may present access and affordability challenges for persons with rare diseases, their families, health care providers, and insurance plans.
- The number of people with one of nine rare diseases in Minnesota varies from 15 living with metachromatic leukodystrophy (MLD) to more than 1,000 living with sickle cell disease (SCD).
- The number of people who may be eligible for gene therapy varies by condition and ranges from fewer than 11 living with beta thalassemia and MLD to as many as 458 living with sickle cell disease.

Background

Rare diseases are complex to manage and frequently have limited approved therapeutic options. The Orphan Drug Act, the U.S. law designed to promote development of drugs for rare diseases, defines a rare disease as one that affects fewer than 200,000 people in the United States.[1] Collectively, rare diseases affect about 25 million people in the United States.[2] Rare diseases often worsen over time and can lead to significant disability, high medical costs, and premature death, sometimes in childhood.

Curative therapies, such as bone marrow transplants, are limited by the frequent lack of suitable donors and the risk of complications associated with utilization of donor bone marrow. One advantage of gene therapies is that they do not require donors. Recent U.S. Food and Drug Administration (FDA) approvals of gene therapies for several rare diseases and additional gene therapies in the pipeline are bringing hope for an improved lifespan and quality-of-life for people with rare diseases.

Many of the currently available gene therapies focus on inherited genetic conditions that manifest during early childhood; however, gene therapies are a growing field and may potentially expand to more common diseases and all age ranges. The gene therapy field is

rapidly evolving and faces several challenges. Challenges include determining appropriate evidence for FDA decisions[3] in the context of small patient populations; limited treatment options and severe outcomes if left untreated; the high cost of individualized treatment; shifting market dynamics[4]; and a shifting regulatory environment.

The rise of gene therapies raises several questions:

- How many people are eligible for gene therapies?
- Among them, who has access to gene therapy treatments?
- What are the anticipated costs for people with rare diseases, their families, and private and public insurance plans?

Data on disease prevalence, including state-level estimates, are needed to address these questions but remain limited.

This analysis focuses on nine rare diseases for which there are FDA-approved gene therapies. The main objective was to estimate the number of people in Minnesota with each disease who may be eligible for these therapies using the Minnesota All Payer Claims Database (MN APCD). These estimates provide a critical first step in projecting the need for and potential costs of these treatments.

Analysis

Nine rare diseases with recently FDA-approved gene therapies (see Table 1) were identified in collaboration with the Minnesota Rare Disease Advisory Council (MNRDAC)[5] and subject matter experts from academia and care delivery.

The analysis included people with one of these nine rare diseases in the MN APCD[6] from 2018 through 2022.

Case definitions, including ICD-10 codes, and criteria for treatment eligibility for gene therapy (age, disease severity, etc.) were derived from the literature with input from subject matter experts. Case numbers and eligibility for gene therapy are based on health insurance claims in the MN APCD.

Table 1 also indicates which diseases are part of the Minnesota Newborn Screening Program, which aims to detect rare diseases that would benefit from early intervention.

Additional details on the analysis methods, including case definitions, can be found in the Supplement—Preparing for the Gene Therapy.

Table 1: Rare diseases with FDA-approved gene therapies included in the analysis

Condition	Part of Minnesota Newborn Screening[7]	Part of Newborn Screening since …	Gene Therapy Proprietary Name	Gene Therapy Nonproprietary Name	Date of FDA Approval
Adrenoleukodystrophy (ALD)[8]	Yes	2/6/2017	Skysona® for cerebral adrenoleukodystrophy (CALD)	elivaldogene autotemcel	9/2022
Beta Thalassemia[9]	No[10]	--	Zynteglo® Casgevy™	betibeglogene autotemcel exagamglogene autotemcel	8/2022 1/2024
Duchenne Muscular Dystrophy (DMD)[11]	Yes	2/24/2025	Elevidys™	delandistrogene moxeparvovec-rokl	6/2023
Dystrophic Epidermolysis Bullosa (DEB)[12]	No	--	Vyjuvek™	beremagene geperpavec-svdt	5/2023
Hemophilia A[13]	No	--	Roctavian™	valoctocogene roxaparvovec-rvox	6/2023[14]
Hemophilia B[15]	No	--	Hemgenix™	etranacogene dezaparvovec-drlb	11/2022
Metachromatic Leukodystrophy (MLD)[16]	Announced in 2025	--	Lenmeldy™	atidarsagene autotemcel	3/2024
Sickle Cell Disease (SCD)[17]	Yes	1988	Casgevy™ Lyfgenia™	exagamglogene autotemcel (exa-cel) lovotibeglogene autotemcel (lovo-cel)	12/2023
Spinal muscular atrophy (SMA)[18]	Yes	3/1/2018	Zolgensma® Itvisma®	onasemnogene abeparvovec-xioi onasemnogene abeparvovec-brve	5/2019 11/2025

Results

The estimated number of people with one of the nine rare diseases in Minnesota during 2018 to 2022 ranged from 15 living with metachromatic leukodystrophy (MLD) to over one thousand living with sickle cell disease (SCD) (Table 2).

Correspondingly, the number of potentially gene therapy-eligible patients in Minnesota varied from fewer than 11 for beta thalassemia and MLD to 458 for SCD.

Table 2: Number of cases and number of cases eligible for gene therapy, 2018-2022

Condition	# Cases	# Treatment-Eligible Cases
Adrenoleukodystrophy (ALD)	121	31
Beta Thalassemia	274	<11
Duchenne Muscular Dystrophy (DMD)	122	119
Dystrophic Epidermolysis Bullosa (DEB)	22	22
Epidermolysis Bullosa Other / Unspecified	122	122
Hemophilia A	656	177
Hemophilia B	191	62
Metachromatic Leukodystrophy (MLD)	15	<11
Sickle Cell Disease (SCD)	1,055	458
Spinal Muscular Atrophy (SMA)	44	38

Source: MDH analysis of MN APCD extract 26.
When reporting data from the MN APCD, MDH follows CMS Cell Size Suppression Policy that any cell (in this case, patient counts) with a value of 1 to 10 shall be suppressed in public reports in order to protect privacy. We denote this by indicating that the count is less than 11.

Conclusions and implications

Results from this analysis indicate a substantial number of Minnesotans with rare diseases are potentially eligible for gene therapies. These therapies could improve the quality of life for people with rare diseases and extend their lives. This creates a need to ensure access to these therapies, including access to specialized medical care and coverage for gene therapies in both public and private insurance plans.

Currently, the direct costs of gene therapies are significant. For example, gene therapies for beta thalassemia have a one-time cost between $2.2 and $2.8 million[19], and gene therapies for sickle cell disease have list prices between $2.2 and $3.1 million.[20] Moreover, access challenges to treatment go beyond the direct cost of the gene therapy. Common barriers for persons with rare diseases include the need to travel to specialized treatment centers, caregiver availability, time off from work or school, housing needs during treatment, insurance coverage for treatment, prior authorization, access to supportive therapies—including fertility preservation and psychosocial support—and the need for comprehensive post-treatment follow-up.

In addition to factors already discussed, the administration of gene therapies requires specialized multidisciplinary teams, clinical capacity, care coordination, and patient navigation. Minnesota is a recognized leader in providing treatment and care for many of the rare diseases mentioned here, and Minnesota-based clinicians provide care to people from other states.

Prevalence data, such as the estimates in this issue brief, are important for planning for appropriate capacity in the healthcare system, as well as insurance coverage. A recent report from the Congressional Budget Office considered the potential impact of gene therapy treatments for SCD on the federal budget. While the report did not go as far as to estimate the budgetary effects of specific policies with regards to gene therapies for SCD, it points to a broad range of potential costs and revenue generation related to the provision of gene therapy and the associated health outcomes.[21] The impacts on the federal budget come from better health, longer life expectancy, higher academic achievement, increased productivity, lower disability rates, and potential savings from lower healthcare costs.

Covering gene therapies are a concern for employers[22] and insurers[23] given the high cost of these therapies and the growing number of gene therapies in development. An analysis by the Employee Benefit Research Institute (EBRI) found that 48 cell and gene therapies had been approved as of 2025, with a growing number targeting more common conditions.[24] While use of these therapies was low in the EBRI analysis of employer-based insurance (0.1% of the enrollee population used a cell or gene therapy), spending for gene therapies accounted for a higher percentage (0.5%) of total spending.

Population estimates provide valuable information to policy makers that may not be available from other sources for forecasting treatment needs and related costs. Although estimates from claims data can be useful for planning, they need to be put into context, acknowledging that not all information to ascertain treatment eligibility is available in claims data.

The MN APCD and other state APCDs can provide valuable estimates given the limitations of other data sources, such as registries, that cover only select populations or electronic health records from a single health system. APCDs can provide an overview of the prevalence of rare diseases and some of the characteristics of the populations that are or will be eligible for gene therapies. As more people access gene therapy treatments covered by public and private health insurance, APCDs will also be able to measure and report prices and spending on these treatments and related care.

Limitations

Treatment of rare disease is specialized care, and it is not uncommon for people to cross state lines to receive treatment. This analysis includes only Minnesota residents. Residents of other states who receive care in Minnesota are not included in this analysis.

Estimates of the number of Minnesotans with each of the nine rare diseases are likely undercounts because the MN APCD lacks reporting in the private insurance market and does not include data for the uninsured or people covered by the Indian Health Service, the Veterans Administration, or TRICARE. Because the type of insurance coverage may vary by condition, change over time, and differ from the general population,[25] the accuracy of the estimated number of cases in this study may also differ by condition. For example, as the MN APCD contains over 95% of Minnesotans covered by public insurance but only about 40% of those

with commercial health insurance, estimates may be more accurate for diseases where most people are covered by public insurance plans (e.g., SCD).

Ascertaining rare disease diagnoses from claims data can be challenging. For instance, ICD-10 diagnosis code G71.01 covers Duchenne and Becker Muscular Dystrophy. To distinguish these two conditions, the Minnesota Department of Health (MDH) applied age restrictions to the case definition (see Supplement—Preparing for the Gene Therapy). Differences in coding, changes in diagnosis codes, and lack of standard case definitions for claims may influence results. To mitigate these effects, algorithms from the literature (when available) and consultations with clinical subject matter experts were used.

Estimates of the number of people eligible for gene therapy treatment for each disease should be interpreted cautiously. Evaluating treatment eligibility for gene therapies requires complex medical decisions. Claims data lack the level of detail available in clinical data and do not capture clinical decision-making informed by laboratory or genetic testing results, disease severity, or the evaluation of different treatment options. As a result, claims analysis can only approximate treatment eligibility and the reported number of eligible people in Table 2 could be over- or underestimated.

For some rare diseases (e.g., hemophilia and SCD) other treatments may be available, influencing treatment decisions about gene therapy. MDH's analysis can only ascertain eligibility based on available data not complex treatment decisions patients make in consultation with their physicians.

Additional disease-specific caveats include the following:

- **Adrenoleukodystrophy (ALD):** Gene therapy for ALD is limited to people with early, active cerebral adrenoleukodystrophy (cALD) which cannot be ascertained from claims. Literature estimates that between 35% and 40% of ALD patients have cALD.[26] Results from newborn screening follow-up in Minnesota indicate that between 2017 and 2022, only two patients developed cALD and both received a stem cell transplant.[27] Therefore, the number of gene therapy-eligible ALD patients here may be an overestimate.
- **Beta Thalassemia**: The number of beta thalassemia cases and gene therapy-eligible transfusion-dependent cases from claims should be considered cautiously. Subject matter experts thought the case count from claims was too high while the number of treatment-eligible patients was too low.
- **Metachromatic Leukodystrophy** (**MLD):** Eligibility for gene therapy is limited to people who are presymptomatic or very early symptomatic. MDH excluded patients with advanced symptoms based on ICD-10 codes from the treatment-eligible population. However, in practice, this requires complex clinical judgment based primarily on motor function and not on medications or procedures that can be found in claims data. Therefore, MDH believes the number of treatment-eligible MLD cases is an overestimation.
- **Sickle Cell Disease** (**SCD)**: People with sickle cell disease are the largest patient group in this analysis and SCD patients are also the largest number of people who are potentially eligible for gene therapy treatment. Not every SCD patient seen in this analysis as potentially

treatment-eligible will be truly treatment-eligible, and some patients may decide on other treatment options (e.g., bone marrow transplant when a donor is available) or to wait for long-term outcome data or future treatments. People with SCD, in particular, face barriers including health insurance coverage, issues with the transition from pediatric to adult care, limited providers with SCD-specific knowledge, stigma, financial barriers, and historical trauma.[28] For these reasons, MDH believes that the number of people with SCD potentially eligible for or pursuing gene therapy may be an overestimation.

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MN APCD
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