

# UNDERSTANDING THE CURRENT LANDSCAPE OF HEALTH INSURANCE COVERAGE FOR RARE DISEASE TREATMENTS

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## What Is Health Insurance?

- Health insurance is a contract that requires a health insurer to pay some, or all your health care costs in exchange for a premium.
- A health insurance contract may also be called a “policy” or “plan”. Your actual document may be called a coverage policy or Summary of Benefits and Coverage (SBC).

## Where Do People Get Health Insurance?

- Through your employer
- Through the Health insurance marketplace (ACA plan)



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# Terminology

**Premium-** The amount that must be paid for your health insurance or plan. You and/or your employer usually pay it monthly, quarterly, or yearly.

**Deductible** – An amount you could owe during a coverage period (usually one year) for covered health care services before your plan begins to pay

**Copay-** A fixed amount (for example, \$15) you pay for a covered health care service, usually when you receive the service.

**Coinsurance** – Your share of the costs of a covered health care service, calculated as a percentage (for example, 20%) of the allowed amount for the service. You generally pay coinsurance plus any deductibles you owe.



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# Terminology

**EOB** - explanation of benefits (EOB) is the insurance company's written explanation regarding a claim, showing what they paid and what the patient must pay. The EOB is not a bill, although it will explain any charges that the patient still owes or may have already paid (in the form of a copay at the time the medical care was received, for example). If the patient owes additional money after the insurance company has paid its portion, the medical provider will send a separate bill, which should match the patient's portion listed on the EOB.

**Claim** - A request for a benefit (including reimbursement of a health care expense) made by you or your health care provider to your health insurer or plan for items or services you think are covered.

**Durable Medical Equipment (DME)** - Equipment and supplies ordered by a health care provider for everyday or extended use. DME may include respiratory equipment, wheelchairs, and orthotics.

**Network** - The facilities, providers and suppliers your health insurer or plan has contracted with to provide health care services.



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# Terminology

**Allowed Amount** – This is the maximum payment the plan will pay for a covered health care service.

**Balance Billing** – When a provider bills you for the balance remaining on the bill that your plan doesn't cover. This amount is the difference between the actual billed amount and the allowed amount.

**Out of Pocket Limit/Maximum Out of pocket** – The most you could pay during a coverage period (usually one year) for your share of the costs of covered services. After you meet this limit, the plan will usually pay 100% of the allowed amount. This limit helps you plan for health care costs. This limit never includes your premium, balance-billed charges or health care your plan doesn't cover. Some plans don't count all your copayments, deductibles, coinsurance payments, out-of-network payments, or other expenses toward this limit.

**Coordination of Benefits (COB)** - A way to figure out who pays first when 2 or more health insurance plans are responsible for paying the same medical claim.



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# Terminology

**Exclusions** - Health care services/treatments/equipment that your plan doesn't pay for or cover

**Denial** - The refusal of an insurance company or carrier to honor a request by an individual (or his or her provider) to pay for health care service, medication, or equipment.

**Peer to Peer** – When your ordering provider speaks with a medical professional representing the insurance plan or reviewing entity

**Grievance** - A complaint about your health insurer

**Appeal** - A request that your health insurer or plan review a decision that denies a benefit or payment (either in whole or in part).

**Prior authorization** - A decision by your health insurer or plan that a health care service is medically necessary. Sometimes called preauthorization, prior approval, PA or precertification. Your health insurance or plan may require prior auth for certain services before you receive them, except in an emergency. PA isn't a promise your health insurance or plan will cover the cost.



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## Employer Sponsored Health Insurance

Many obtain commercial coverage through their employer.

Technically, no business has to offer health insurance to their employees. However, under the Affordable Care Act (ACA), larger businesses with 50+ FTE employees will receive a dollar amount tax penalty per employee if they do not offer health insurance.

Some employer plans have grandfathered status and are exempt from certain ACA rules.

According to Kaiser Family Foundation, **67%** of covered workers in America were enrolled in a self-insured plan in 2025.

Employers that offer health insurance benefits finance those benefits in one of two ways.

- **Self-funded** (also referred to as “ERISA” plans)
- **Fully insured**



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## Self Insured = Federally Regulated

- Many employers choose to 'self-insure' rather than purchase health insurance for their employees. This means the employer pays the health care claims for all employees.
- The U.S. Department of Labor generally provides oversight of self-insured employer plans. Other agencies, including the Office of Personnel Management, the Defense Health Agency, and the Centers for Medicare and Medicaid Services, provide oversight when the employer is a government agency.
- These plans have less patient protections than fully insured plans given that employers are permitted to “laser” out specific diagnoses and/or approved treatments.
- Most contract with traditional health plans to administer their claims, prior authorizations, etc. The health plans acts as a Third Party Administrator (TPA).



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## Financial Tools Employers Use for Risk Mitigation

- Stop Loss-(also known as excess insurance) is a product that provides protection against catastrophic or unpredictable losses.
- Specific-provides protection for the employer against a high claim on any one individual.
- Aggregate- provides a ceiling on the dollar amount of eligible expenses that an employer would pay, in total, during a contract period.
- Reinsurance-is effectively "insurance for insurance companies." When a health plan covers an individual with extremely expensive medical needs, the reinsurance policy pays a portion of that claim.

<https://www.hcaa.org/page/selffundingstoploss>



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# Fully Insured = State Regulated

- Fully insured employer health plans are regulated and overseen by each state. Each state has an insurance commission which regulates the business of insurance.

## **Key Differences of Fully Insured Plans:**

- **Risk & Costs:** Fully insured plans have fixed, predictable monthly costs. Self-insured plans have variable costs, where employers assume the risk of high-claims years but save on premiums in low-claim years.
- **Administration & Flexibility:** Fully insured plans are standardized, allowing limited customization. Self-insured plans are highly customizable to meet specific workforce needs.
- **Regulation:** Fully insured plans are subject to state regulations, while self-insured plans are mostly governed by federal law (ERISA).



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## ACA Marketplace Plans

- The federal government operates the Health Insurance Marketplace®, available at [HealthCare.gov](https://www.healthcare.gov), for most.
- Some states run their own Marketplaces.
- Open Enrollment runs from Nov 1 – Jan 15.
- Navigators are provided to help figure out which plan may work best for individuals
- Marketplace plans are private plans that must be preapproved by each state insurance department before they are sold on the marketplace to their state's residents



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# ACA Marketplace Plans-Essential Health Benefits Rule

Per the ACA, these 10 essential health benefits must be covered: All plans offered in the Marketplace cover these 10 essential health benefits:

Ambulatory patient services (outpatient care you get without being admitted to a hospital)

Emergency services

Hospitalization (like surgery and overnight stays)

Pregnancy, maternity, and newborn care (both before and after birth)

Mental health and substance use disorder services, including behavioral health treatment (this includes counseling and psychotherapy)

Prescription drugs

Rehabilitative and habilitative services and devices (services and devices to help people with injuries, disabilities, or chronic conditions gain or recover mental and physical skills)

Laboratory services

Preventive and wellness services and chronic disease management

Pediatric services, including oral and vision care (but adult dental and vision coverage aren't essential health benefits)

Plans **must** also include the following benefits:

Birth control coverage

Breastfeeding coverage

Essential health benefits are minimum requirements for all Marketplace plans. Specific services covered in each broad benefit category can vary based on your state's requirements.



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## Medicaid

- Largest source of health coverage in the United States
- Administered by states per federal requirements
- Jointly funded by both the states and the federal government

## Eligibility

- Low-income families
- Qualified pregnant women and children
- Individuals receiving SSI (supplemental security income)
- Other groups as determined by Medicaid agency –disabled

**Important: appeals of a Medicaid denial are resolved by requesting a Fair Hearing. You can also have an advocate, such as your provider, present with you**



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## Patient Protections Unique to Medicaid

- **EPSDT (Early and Periodic Screening, Diagnostic, and Treatment)**, is a comprehensive Medicaid benefit for children under 21 that ensures access to necessary preventive, dental, mental health, and specialty services, aiming to identify and treat health conditions early to improve children's health.
- **MDRP (Manufacturer's Drug Rebate Program)** requires pharmaceutical manufacturers to pay rebates to Medicaid on outpatient drugs, reducing costs for state Medicaid programs. It ensures Medicaid receives the lowest available prices for prescription drugs. Medicaid has to cover drugs on the MDRP regardless of approval pathway; however, Medicaid programs are allowed to develop prior authorization criteria.
- Medicaid programs perform clinical reviews on all newly approved treatments via **DUR/P&T (Drug Utilization Review/Pharmacy & Therapeutics)** Committees made up of healthcare providers. Unlike private health plans, these meetings are public giving patient advocacy organizations an opportunity to provide comment on any proposed criteria prior to final decision, however, these committees lack rare disease expertise.



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## So what does all this mean for rare disease patients? Our experience shows:

- Most health insurers do not cover treatments approved via FDA's Accelerated Approval pathway, deeming them “investigational/experimental” resulting in a denial.
- If they do cover, they will sometimes issue prior authorization criteria that limits coverage to what the clinical trial population looked like which is normally very restrictive in comparison to the FDA label. Any patient falling outside of their criteria will receive a denial.
- Health plans decide whether a drug will be covered under their medical or pharmaceutical benefit, which can greatly impact a patient's out of pocket costs.
- Self insured employers do not have to adhere to the ACA's Essential Health Benefits rule. SIE's have the authority to exclude coverage for specific treatments (all cell/gene therapies), services (organ transplants), etc. If the plan document specifically excludes it, patient will receive a denial regardless of medical necessity.



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# Coverage Policies – Forzinity (Accelerated Approval)

Cigna:

**Forzinity® (elamipretide subcutaneous injection) is(are) covered as medically necessary when the following criteria is(are) met for FDA-approved indication(s) or other uses with supportive evidence (if applicable):**

## **FDA-Approved Indication**

- 1. Barth Syndrome.** Approve for 1 year if the patient meets ONE of the following (A or B):
  - A) Initial Therapy.** Approve if the patient meets ALL of the following (i, ii, and iii):
    - i.** The patient weighs  $\geq$  30 kg; AND
    - ii.** The diagnosis is established by ONE of the following (a, b, or c):
      - a)** Patient has a laboratory test demonstrating an increased ratio of monolysocardiolipin (MLCL)/cardiolipin (CL) on erythrocytes, tissue, fibroblasts, or stored neonatal bloodspots; OR
      - b)** Patient has a laboratory test demonstrating elevated 3-methylglutaric acid, 3-methylglutaconic acid (3-MGC), and 2-ethylhydracrylic acid on urine organic acids analysis; OR
      - c)** Patient has a molecular genetic test demonstrating a hemizygous pathogenic variant in the tafazzin (TAZ) gene; AND
    - iii.** The medication is prescribed by or in consultation with a geneticist, cardiologist, metabolic specialist, hematologist, pediatrician, or a physician who specializes in the treatment of mitochondrial disorders; OR
  - B) Patient is Currently Receiving Forzinity.** Approve if the patients meets BOTH of the following (i and ii):
    - i.** Patient has been established on therapy for at least 1 year; AND  
Note: A patient who has received < 1 year of therapy or who is restarting therapy with the requested drug should be considered under criterion A (Initial Therapy).
    - ii.** According to the prescriber, the patient has demonstrated a clinical response, defined as stabilization or lack of decline from baseline (prior to initiating Forzinity).  
Note: Examples of a clinical response include stabilization or lack of decline in muscle strength, balance, six-minute walking distance, or fatigue.



# Coverage Policy – Kygevvvi (Standard Approval)

Ambetter Health (Centene subsidiary that provides Marketplace plans:

It is the policy of health plans affiliated with Centene Corporation® that Kygevvvi is **medically necessary** when the following criteria are met:

## I. Initial Approval Criteria

### A. Thymidine Kinase 2 Deficiency (must meet all):

1. Diagnosis of TK2d as evidenced by a mutation in the thymidine kinase
2. Prescribed by or in consultation with a neurologist or metabolic disease
3. Age of symptom onset  $\leq$  12 years (e.g., proximal muscle weakness, respiratory weakness, facial diplegia; *see Appendix D*);
4. Documentation of member's current weight in kg;
5. Dose does not exceed doxycitine 400 mg/kg and doxorubicin 400 mg/kg (*see Appendix E for the recommended number of packets per day based on dose*).

**Approval duration: 12 months**

### B. Other diagnoses/indications (must meet 1 or 2):

1. If this drug has recently (within the last 6 months) undergone a label change (newly approved indication, age expansion, new dosing regimen) that reflected in this policy, refer to one of the following policies (a or b):
  - a. For drugs on the formulary (commercial, health insurance marketplace, Medicaid), the no coverage criteria policy for the relevant line of business: CP.CPA.190 for commercial, HIM.PA.33 for health insurance marketplace, and CP.PMN.255 for Medicaid; or
  - b. For drugs NOT on the formulary (commercial, health insurance marketplace, Medicaid), the non-formulary policy for the relevant line of

CP.CPA.190 for commercial, HIM.PA.103 for health insurance marketplace, and CP.PMN.16 for Medicaid; or

2. If the requested use (e.g., diagnosis, age, dosing regimen) is NOT specifically listed under section III (Diagnoses/Indications for which coverage is NOT authorized) AND criterion 1 above does not apply, refer to the off-label use policy for the relevant line of business: CP.CPA.09 for commercial, HIM.PA.154 for health insurance marketplace, and CP.PMN.53 for Medicaid.

## II. Continued Therapy

### A. Thymidine Kinase 2 Deficiency (must meet all):

1. Member meets one of the following (a or b):
  - a. Currently receiving medication via Centene benefit or member has previously met initial approval criteria;
  - b. Member is currently receiving medication and is enrolled in a state and product with continuity of care regulations (*refer to state specific addendums for CC.PHARM.03A and CC.PHARM.03B*);
2. Member is responding positively to therapy;
3. Documentation of member's current weight in kg;
4. If request is for a dose increase, new dose does not exceed doxycitine 400 mg/kg and doxorubicin 400 mg/kg per day (*see Appendix E for the recommended number of packets per day based on total daily dose*).

**Approval duration: 12 months**



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# Self Insured Plan Document-Exclusions



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## Denials

- Of the reasons for receiving a denial, two major ones are 1) Health plan deems it not medically necessary, investigational or experimental, or 2) Claim is for something simply not covered by the plan.
- ❖ Medical Necessity: internal appeals process is determined by the plan document, and all fully insured plans are required to have an internal appeals process, including using Independent Review Organizations (IRO's) when all internal plan appeals are exhausted. IRO's are external reviewers that have no self interest in the outcome.
- ❖ Coverage Dispute: complaints over whether something is covered under the health plan are overseen by state insurance regulators, since these complaints involve the language of the health plan which is based in contract law. Note: all state regulated health plans have to be pre-approved by each state insurance commission before they can be sold in that particular state.



# Appeal Letter

Member Name: [REDACTED]

ID Number: [REDACTED]

DOB: [REDACTED]

Services: Medication Administration J1413; 36415; 96363; 96366; 96375

To whom it may concern:

I am writing to appeal and request an overturn on the most recently denied request for Elevidys for my son, [REDACTED]. I believe that your decision to deny is wrong, based on the fact this treatment is medically necessary. Elevidys is an FDA approved treatment for individuals 4 years of age and older with DMD with a confirmed mutation in the DMD gene. [REDACTED] has completed all the screenings and the required testing to confirm that he is eligible for this treatment. This is an urgent and time-sensitive request, and we are requesting an expedited review for medical necessity by a neuromuscular specialist with experience treating patients with Duchenne.

[REDACTED] is an amazing little boy. *Talk about him here and who he is/what he loves. Discuss his independence/skills that are important that he maintain. Mention how [REDACTED] and your family are affected (including your other son who has DMD and has received treatment).*

Elevidys received FDA approval on June 20, 2024 in individuals 4 years of age and older with DMD with a confirmed mutation in the DMD gene, meeting FDA's standard for safety and effectiveness that all FDA-approved medicines must meet. The criterion for denial stated by the BlueCross BlueShield Federal Employee Program is that patients can only be approved for this treatment if there is NO deletion in exons 1 to 17 and/or exons 59 to 71 in the DMD gene. This simply does not align with the FDA label. Page two of the policy itself indicates that the only mutations that are contraindicated for this treatment are deletions in exon 8 and/or exon 9 in the DMD gene. My son has no mutation in that area. My son's clinical team is aware of where [REDACTED]'s mutation is and has determined that this treatment is medically necessary. Based on where my son's mutation is, they are fully aware that there could be a chance of additional risk. Included in the guidance is the consideration of additional immunomodulatory treatment if symptoms do occur. This potential risk does not change the medical necessity of this treatment. In addition, [REDACTED]'s brother (who has the same mutation) has already received this treatment and at the time of his treatment the plan covered him without denials. I don't know how this treatment could be deemed by the plan to be medically necessary for one of my children, but not the other when they have the same disease and mutation.

Per the Brochure:

*All benefits are subject to the definitions, limitations, and exclusions in this brochure and are payable only when we determine that the criteria for medical necessity are met. Medical necessity shall mean healthcare services that a physician, hospital, or other covered professional or facility provider, exercising prudent clinical judgment, would provide to a patient for the purpose of preventing, evaluating, diagnosing, or treating an illness, injury, disease, or its symptoms, and that are:*

1. In accordance with generally accepted standards of medical practice in the United States; and  
2. Clinically appropriate, in terms of type, frequency, extent, site, and duration; and considered effective for the patient's illness, injury, disease, or its symptoms; and

3. Not primarily for the convenience of the patient, physician, or other healthcare provider, and not more costly than an alternative service or sequence of services at least as likely to produce equivalent therapeutic or diagnostic results for the diagnosis or treatment of that patient's illness, injury, or disease, or its symptoms; and

4. Not part of or associated with scholastic education or vocational training of the patient; and

5. In the case of inpatient care, able to be provided safely only in the inpatient setting.

For these purposes, "generally accepted standards of medical practice" means standards that are based on credible scientific evidence published in peer-reviewed medical literature generally recognized by the relevant medical community and physician specialty society recommendations.

The experts at FDA have determined that Elevidys is safe and effective. I would further like to point out that Elevidys is listed in accepted Compendia including Wolters Kluwer's Lexi-drugs with no question of safety and indications for usage. There is multiple peer-reviewed scientific literature published in support of Elevidys. Elevidys received orphan drug designation in 2018 for treatment of Duchenne Muscular Dystrophy. It is clinically appropriate. His medical team believes this treatment is necessary to treat his Duchenne. There is ample documentation available to show that Elevidys is effective in the treatment of DMD and there is no reason that it should not be covered for [REDACTED]. There are no alternative service or services that are at least as likely to produce equivalent results as this gene therapy. Therefore, treatment meets medical necessity per the brochure.

If you have additional questions, you can reach out to my son's Neuromuscular Clinic at the Arkansas Children's Hospital or myself, at *(insert preferred contact here)*.

Thank you for your consideration.

Sincerely,

[REDACTED]

[REDACTED]'s mother)



# Checklist:

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- ❑ Obtain and read a copy of your **plan document**. These can be lengthy so pay particular attention to the sections “Exclusions/Limitations” and “Appeals.”
- ❑ Let your provider know that if they receive a denial, to let you know as soon as possible. Health insurers are required to send you copies of all denials and the reason for denial, but providers usually receive them before patients. Expedited review can be requested and granted if it meets certain conditions.
- ❑ Maintain copies of any and all denials. These contain important deadlines as well as instructions on how to appeal the decision.
- ❑ See the process through to External Review. Make sure you and your provider are coordinating on this. Always request an External Review based on medical necessity.
- ❑ If you feel as if your health insurer improperly issued a denial, you can file a complaint. For self-insured employer plans: [DOL-ERISA](#). For fully-insured plans you can find your state regulator here: [National Association of Insurance Commissioners](#). For Marketplace plans: [Marketplace Appeals](#). For Medicaid, request a fair hearing--instructions on how to do this will be on the Notice of Adverse Determination you/your provider receive.

