Office of Medicaid
BOARD OF HEARINGS

Appellant Name and Address:

[redacted]

Appeal Decision:	Denied	**Appeal Number:**	2504366
Decision Date:	5/5/2025	**Hearing Date:**	4/23/2025
Hearing Officer:	David Jacobs		

Appearance for Appellant:
[redacted]

Appearances for MCO:
Jacqueline Bigbee, Director of Member Appeals & Grievances
Felicia DiSciscio, Manager of Member Appeals & Grievances
Dr. William Keough, Senior Medical Director
Dr. Heather Schlott, Physician Reviewer
Priya Mehta, Assistant General Counsel

The Commonwealth of Massachusetts
Executive Office of Health and Human Services
Office of Medicaid
Board of Hearings
100 Hancock Street, Quincy, Massachusetts 02171

APPEAL DECISION

Appeal Decision:	Denied	**Issue:**	Managed Care Organization; Denial of Internal Appeal; Genetic Testing
Decision Date:	5/5/2025	**Hearing Date:**	4/23/2025
MCO Reps.:	Jacqueline Bigbee, Felicia DiSciscio, Dr. William Keough, Dr. Heather Schlott, Priya Mehta	**Appellant's Reps.:**	[redacted]
Hearing Location:	Telephonic	**Aid Pending:**	No

Authority

This hearing was conducted pursuant to Massachusetts General Laws Chapter 118E, Chapter 30A, and the rules and regulations promulgated thereunder.

Jurisdiction

Through a letter dated February 6, 2025, Wellsense Health Plan ("Wellsense"), a MassHealth managed care organization ("MCO"), denied the appellant's internal Standard Appeal for coverage of genetic testing because it is not medically necessary (Exhibit 1). The appellant filed this appeal in a timely manner on March 18, 2025 (Exhibit 2). Denial of authorization by a managed care contractor is valid grounds for appeal (130 CMR 610.032(B).)

Action Taken by the MCO

Wellsense denied the appellant's request for genetic testing because it is not medically necessary.

Issue

The appeal issue is whether Wellsense was correct, pursuant to 130 CMR 450.204, in determining that the appellant's requested procedure is not medically necessary.

Summary of Evidence

The appellant is a MassHealth member who is enrolled in Wellsense Health Plan, a MassHealth MCO. Both parties appeared telephonically via Zoom. Pg. 69 of the appeal packet summarizes the issue on appeal as follows:

"[The Appellant] is a [redacted] who presents in Genetics Clinic at RIH/HCH CNDC for evaluation due to his diagnosis of type 1 diabetes and concurrent exocrine pancreatic insufficiency, suggesting a dual endocrine and exocrine pancreatic dysfunction. CFTR related disorders would be on the differential, but unlikely in the setting of normal newborn screening and normal sweat test (sweat chloride 21/22 mmol). He has normal developmental milestones and is non dysmorphic on exam today. The upcoming MRCP will be helpful to determine if structural abnormalities are contributing. We would also recommend exome sequencing (ES), which will evaluate for a broad range of genetic conditions that could underlie [the appellant's] pancreatic dysfunction, including monogenic diabetes, hereditary pancreatitis (PRSS1, SPINK, CTRC, CLDN2, CPA1, CASR, GEL. TRPV6), genetic syndromes (Johanson-Blizzard, Shwachman-Diamond or Pearson marrow-pancreas, IPEX, amongst others) or rare CFTR-related mutations. Exome sequencing is preferable over a targeted panel because it offers a comprehensive evaluation, allowing for the detection of both known and potentially novel variants that might not be included in a pancreas-specific gene panel, thereby maximizing the diagnostic yield."

(Exhibit 5, pg. 69).

After an initial denial of the request for prior authorization of exome sequencing, an internal appeal was filed with Wellsense on January 8, 2025. The internal appeal was denied on February 6, 2025, which is the notice on appeal today (Exhibit 1). The notice summarizes the reasoning for denial of exome sequencing as follows: "your child's request for genetic testing is considered not medically necessary for the treatment of problem of the pancreas because causes of the problem not related to [the appellant's] genes have not been eliminated. Additionally, there is no documentation that the results of the requested test will change [the appellant's] plan of care. This decision is based on EviCore criteria for Exome Sequencing, Mitochondrial Disorders Genetic Testing Policy" (Exhibit 1).

The appellant's request describes exome sequencing and why it is preferable to them than other diagnostic methods as follows:

"Exome sequencing (ES) uses next generation sequencing technology is the most effective method to simultaneously analyze the majority of the genes in the genome. Published data from clinical exome tests indicate a diagnostic yield of greater than 30%, which is significantly higher than many phenotype-specific panels. It has been suggested that ES be pursued as the next step following a negative phenotype-driven next generation sequencing panel for increased detection rates. In a large cohort of 500 ES cases 7.5% were found to have a novel gene finding

that would not have been identified by clinically available phenotype-driven panels. ES is the most cost-effective and comprehensive method to rapidly detect the underlying cause in patients afflicted with genetic disease, as 85% of genetic mutations with large clinical consequences occur within the exome. A 2016 study by Monroe supports the cost-effectiveness of ES. In this study the average cost of a diagnostic odyssey using non-ES techniques yielded an average cost of $16,409 per patient. ES identified a diagnostic result in 29.4% of patients tested. Trio-ES saved an average of $3,547 per patient with a diagnosis and $1,727 for undiagnosed patients. Thus making ES a cost-effective diagnostic tool."

(Exhibit 5, pg. 69).

At the hearing, Dr. Heather Schlott and Dr. William Keough spoke on behalf of Wellsense. [redacted] represented the appellant. Dr. Schlott testified that the reason the appellant's request was denied is because it does not meet Wellsense's criteria for genetic testing for both mitochondrial testing and exome sequencing. Wellsense puts special emphasis on the fact that these criteria are "and" criteria, meaning all bullet points must be met for a finding of medical necessity.

For mitochondrial testing, the relevant criteria for coverage per the Wellsense handbook are as follows:

Diagnostic Testing for Symptomatic Individuals:

- **Member has multiple organ system involvement defined as altered function in two or more organ systems, suggestive of a mitochondrial disorder, and**
- Member has one or more of the following clinical features: proximal myopathy, cardiomyopathy, encephalopathy, seizures, dementia, stroke-like episodes, ataxia, spasticity, ptosis, ophthalmoparesis, ophthalmoplegia, optic atrophy, pigmentary retinopathy, sensorineural hearing loss, diabetes mellitus, mid- or late pregnancy loss, brain magnetic resonance imaging (MRI) or magnetic resonance spectroscopy (MRS) results consistent with a mitochondrial process, and/or pathology results consistent with a mitochondrial process, and
- Targeted mutation analysis is not feasible because of one of the following:
    - Member's clinical presentation does not fit a well-described syndrome for which single-gene or targeted panel testing is available (see table titled *Select Mitochondrial Disorders*), or
    - Member's clinical presentation fits a well-described syndrome and applicable single-gene or targeted mutation analysis was negative, and
- Alternate etiologies have been considered and ruled out when possible (e.g., environmental exposure, injury, infection), and
- Family history strongly suggests mitochondrial inheritance (e.g., no evidence of paternal transmission), AND

(Exhibit 5, pg. 111) (emphasis added).

Wellsense concedes that the appellant meets the second bullet point but noted that he meets none of the others. Primarily, he does not meet the first criteria as the only organ at issue is his pancreas.

The appellant's representative responded that the appellant's unusual combination of endocrine and exogen insufficiency suggests that there may be other organs involved, and other genetic issues involved that would allow the appellant to meet the criteria. However, Wellsense disagreed with that argument, noting that problems with just the pancreas can account for the insufficiencies she notes.

For exome sequencing, the relevant criteria for coverage per the Wellsense handbook are as follows:

"A genetic etiology is considered the most likely explanation for the phenotype, based on ONE of the following:

- Unexplained epileptic encephalopathy (onset before three years of age) and no prior epilepsy multigene panel testing performed, OR
- Global developmental delay (significant delay in younger children, under age 5 years, in at least two of the major developmental domains: gross or fine motor; speech and language; cognition; social and personal development; and activities of daily living) following formal assessment by a developmental pediatrician or neurologist, OR
- Moderate/severe/profound intellectual disability (defined by Diagnostic and Statistical Manual of Mental Disorders [DSM-5] criteria, diagnosed by 18 years of age) following formal assessment by a developmental pediatrician or neurologist, OR
- Multiple congenital abnormalities defined by ONE of the following:
    - Two or more major anomalies affecting different organ systems*, or
    - One major and two or more minor anomalies affecting different organ systems, OR
- TWO of the following criteria are met:
    - major abnormality affecting at minimum a single organ system*, and/or
    - formal diagnosis of autism, and/or
    - symptoms of a complex neurodevelopmental disorder (e.g., self-injurious behavior, reverse sleep-wake cycles, dystonia, ataxia, alternating hemiplegia, neuromuscular disorder), and/or
    - severe neuropsychiatric condition (e.g., schizophrenia, bipolar disorder, Tourette syndrome), and/or
    - period of unexplained developmental regression, and/or
    - laboratory findings suggestive of an inborn error of metabolism, AND

- **Alternate etiologies have been considered and ruled out when possible (e.g., environmental exposure, injury, infection),** AND
- Clinical presentation does not fit a well-described syndrome for which first tier testing (e.g., single gene testing, comparative genomic hybridization [CGH]/chromosomal microarray analysis [CMA]) is available, AND
- Multiple targeted panels are appropriate based on the member's clinical
- presentation, AND
- **There is a predicted impact on health outcomes including:**
    - **Application of specific treatments**, or
    - Withholding of contraindicated treatments, or
    - Surveillance for later-onset comorbidities, or
    - Initiation of palliative care, or
    - Withdrawal of care, AND
- A diagnosis cannot be made by standard clinical work-up, excluding invasive procedures such as muscle biopsy, AND
- Rendering laboratory is a qualified provider of service per the Health Plan policy.

(Exhibit 5, pgs. 93-94) (Emphasis added).

Wellsense found that the appellant did not meet the criteria for exome sequencing on three primary grounds. First, the appellant does not have the required developmental delays or cognitive abnormalities necessary to meet the criteria. Second, non-genetic causes such as a biochemical cause in the pancreas have not been ruled out to meet the criteria. Lastly, there is no proof that treatment of the appellant's condition would change if exome sequencing were performed. Wellsense argues that there are less costly alternatives available to the appellant in the form of panels for individual conditions that the appellant must try before exome sequencing may be covered.

The appellant's representative conceded that the appellant does not meet these criteria. However, she argues these criteria are not best medical practices as the appellant is presenting an unusual combination of endocrine and exogen insufficiency, suggesting that his condition is related to genetics and best diagnosed with genetic testing. She emphasized that he does not have any development delays now, but they may arise in the future due to the unusual insufficiencies he is exhibiting. Such conditions are best caught early, which is best done with genetic testing. The length of time it will take to do individual panels may be detrimental to the appellant's health. Moreover, the cost of many individual panels is likely to exceed the cost of genetic testing, making exome sequencing the least costly alternative.

In response, Wellsense argued that they do not believe that exome sequencing is more likely to be a more efficient, less costly testing method than individual panels. They argue there are always more thorough tests that can be done. Moreover, the Wellsense doctors do not agree that the endocrine and exogen insufficiency exhibited by the appellant necessarily means he

has a more complex condition than what is apparent in the pancreas. A bio-chemical problem in the pancreas may explain all the appellant's symptoms. Regarding the argument that the criteria is not best medical practices, Wellsense argued that these criteria have been developed by geneticists, submitted to MassHealth, and approved as a satisfactory means to find the least costly alternatives for medical necessity. They argued that "the criteria are the criteria" for medical necessity in accordance with the MassHealth regulations. and the appellant does not meet them. Furthermore, Wellsense is not willing to make any special exceptions for the appellant, as this is only done when there is a close call or no other test available, and that is not the situation here.

Findings of Fact

Based on a preponderance of the evidence, I find the following:

1) The appellant is a MassHealth member who is enrolled with Wellsense Health Plan, a managed care organization administering MassHealth benefits.

2) The appellant is a minor male child with a diagnosis of type 1 diabetes and concurrent exocrine pancreatic insufficiency, suggesting a dual endocrine and exocrine pancreatic dysfunction.

3) The appellant submitted a prior authorization request for exome sequencing that was denied by Wellsense, and an internal appeal was filed on January 8, 2025.

4) On February 6, 2025, Wellsense denied the internal appeal.

5) The appellant appealed the February 6, 2025 denial to the Board of Hearings.

6) The pancreas is the only organ of the appellant that shows signs of dysfunction.

7) It is not clear if the appellant's endocrine and exocrine pancreatic dysfunction has a genetic cause.

8) The appellant exhibits no developmental delays or cognitive abnormalities.

9) Non-genetic causes for the appellant's condition have not been ruled out.

10) No alternative treatment plan for a genetic cause was submitted.

Analysis and Conclusions of Law

MassHealth members who do not have another form of insurance are generally enrolled in a managed care organization. (See 130 CMR 508.001; 508.002; see also 130 CMR 450.105.) Managed care organizations provide "management of medical care, including primary care, behavioral health services, and other medical services" for enrolled members. (130 CMR 450.117(B).) Members enrolled in a managed care provider are entitled to a fair hearing under 130 CMR 610.000 to appeal a determination by a managed care organization if the member has exhausted all remedies available through the contractor's internal appeal process. (130 CMR 508.010.)

MassHealth and MCOs cover only those services that are deemed "medically necessary." The MassHealth regulations define a "medical necessity" service as follows:

> (1) it is reasonably calculated to prevent, diagnose, prevent the worsening of, alleviate, correct, or cure conditions in the member that endanger life, cause suffering or pain, cause physical deformity or malfunction, threaten to cause or to aggravate a handicap, or result in illness or infirmity; and
>
> (2) there is no other medical service or site of service, comparable in effect, available, and suitable for the member requesting the service, that is more conservative or less costly to the MassHealth agency. Services that are less costly to the MassHealth agency include, but are not limited to, health care reasonably known by the provider, or identified by the MassHealth agency pursuant to a prior-authorization request, to be available to the member through sources described in 130 CMR 450.317(C), 503.007: *Potential Sources of Health Care*, or 517.007: *Utilization of Potential Benefits*.

130 CMR 450.204(A).

Here, Wellsense has set forth its criteria for determining the medical necessity of mitochondrial testing and exome sequencing in the Wellsense handbook (Exhibit 5, pgs. 109-113 and pgs. 92-95). Wellsense persuasively argued that the criteria are non-arbitrary standards, created by medical professionals, and accepted by MassHealth as an objective method by which to evaluate medical necessity.

The appellant representative concedes that the appellant does not meet the Wellsense criteria but argues that the criteria themselves are not best medical practices. She argues that the cost of the exome sequencing is equivalent or close to the individual panels that Wellsense suggests is the next step for the appellant. She argues that the multitude of individual panels that will be required will take longer and be more costly than exome sequencing which will do the work of several

panels all at once. Moreover, she argues that the endocrine and exocrine pancreatic dysfunction that the appellant exhibits may be early warning signs of the kind of genetic developmental delays and cognitive dysfunction that would eventually lead to the appellant meeting the other criteria. She argues that delaying diagnoses of these conditions will be to the appellant's detriment.

Wellsense disagrees with the appellant's position that exome sequencing would be less expensive than individual panels, as there are always more costly tests that can be done when exploring the cause of a condition. The Wellsense doctors also disagree that endocrine and exocrine pancreatic dysfunction necessarily means the appellant has a genetic disorder as it may also have a bio-chemical cause within the pancreas. Wellsense believes that panel screening will be the most effective, least costly method of diagnosing the appellant where he shows no genetic causes for his condition based on their criteria for genetic testing.

Notwithstanding the appellant's argument above, it is undisputed that the appellant does not meet the medical necessity criteria that Wellsense has established. The parties' opposing positions regarding whether exome sequencing is the best next step for the appellant is, ultimately, not determinative here. The appellant's representative has not presented any objective evidence to suggest that the Wellsense criteria for exome sequencing need not be followed. The appellant's argument that following the Wellsense protocol *may* result in a less efficient diagnoses process that *may* lead to a worsening of his condition falls short of meeting the appellant's burden here. The appellant has not demonstrated that Wellsense erred in its determination that he has not shown that the requested service is medically necessary.

For these reasons, this appeal is DENIED.

Order for Wellsense Health Plan

None.

Notification of Your Right to Appeal to Court

If you disagree with this decision, you have the right to appeal to Court in accordance with Chapter 30A of the Massachusetts General Laws. To appeal, you must file a complaint with the Superior Court for the county where you reside, or Suffolk County Superior Court, within 30 days of your receipt of this decision.

David Jacobs
Hearing Officer
Board of Hearings

cc:

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