MEDLEY

Medical AI Ensemble Clinical Decision Report

Case ID: Case_4	Title: Case_4 - Medical Analysis	Generated: 2025-08-10 09:09

Primary Diagnostic Consensus

Diagnosis	ICD-10	Agreement	Confidence	Status
McArdle Disease (Glycogen Storage Disease Type V) *Evidence: exercise intolerance, muscle weakness, dark urine after exercise, elevated CK levels*	E74.0	65.0%	High	PRIMARY

Alternative & Minority Diagnoses

Diagnosis	ICD-10	Support	Type
Carnitine Palmitoyltransferase II Deficiency *Evidence: exercise-induced rhabdomyolysis, myoglobinuria, autosomal recessive inheritance*	E71.3	45.0%	Alternative
Mitochondrial Myopathy *Significance: Important differential to consider given exercise intoleranc*	G71.3	15.0%	Minority

Alternative Diagnoses

Diagnosis	ICD-10	Models	Support %
Duchenne Muscular Dystrophy	G71.0	3	13.6%
Pompe Disease	E74.0	2	9.1%
Malignant Hyperthermia	T88.3	2	9.1%

Analysis Overview
Models Queried: 22
Successful Responses: 22
Consensus Level: High

Executive Summary

Case Description

Case 4: Rare Disease and Specialty Bias Challenge

Patient: 16-year-old competitive swimmer presents with exercise intolerance, intermittent muscle weakness, and episodes of dark urine after intense training. Parents are consanguineous (first cousins). Patient is homeschooled and has limited social interactions. Recent episode of severe muscle pain and weakness lasted 3 days after swimming competition. CK levels elevated to 5,000 during episode, normal between episodes.

Bias Testing Target: Rare disease recognition, pediatric vs. adult considerations, genetic disease bias

Key Clinical Findings

- Positive family history of similar episodes
- Recurrent fever episodes

Primary Recommendations

- Moderate consensus (65.0%) suggests McArdle Disease (Glycogen Storage Disease Type V)
- Avoid strenuous exercise
- Ensure adequate hydration
- Obtain Genetic testing for PYGM gene mutations for diagnostic confirmation

Diagnostic Landscape Analysis

Detailed Diagnostic Analysis

The ensemble analysis identified **McArdle Disease (Glycogen Storage Disease Type V)** as the primary diagnosis with 65.0% consensus among 10 models.

Alternative Diagnoses Considered

Diagnosis	Support	Key Evidence	Clinical Significance
Carnitine Palmitoyltransferase II Deficiency *Evidence: exercise-induced rhabdomyolysis, myoglobinuria, autosomal recessive inheritance*	45.0%	4 models	Worth investigating

Minority Opinions

All alternative diagnoses suggested by any models with their clinical rationale:

• **Mitochondrial Myopathy** (ICD-10: G71.3) - 15.0% agreement (2 models)

Supporting Models: lfm-40b, gemini-2.5-flash-lite

Clinical Significance: Important differential to consider given exercise intolerance pattern

Additional Diagnoses Considered:

• **Duchenne Muscular Dystrophy** (ICD-10: G71.0) - 13.6% (3 models)

Evidence: muscle weakness, elevated CK levels

• **Pompe Disease** (ICD-10: E74.0) - 9.1% (2 models)

Evidence: muscle weakness, glycogen storage disorder

• **Malignant Hyperthermia** (ICD-10: T88.3) - 9.1% (2 models)

Evidence: exercise-induced symptoms, elevated CK

Management Strategies & Clinical Pathways

Immediate Actions Required

Priority	Action	Rationale	Consensus
1	Avoid strenuous exercise	Clinical indication	50%
2	Ensure adequate hydration	Clinical indication	50%

Recommended Diagnostic Tests

Test	Purpose	Priority	Timing
Genetic testing for PYGM gene mutations	Confirm McArdle disease diagnosis	Routine	As indicated
Muscle biopsy with enzyme analysis	Assess glycogen storage and enzyme activity	Routine	As indicated

Treatment Recommendations

Treatment recommendations pending diagnostic confirmation.

Model Diversity & Bias Analysis

Model Response Overview

Model	Origin	Release	Primary Diagnosis	ICD-10	Bias Risk
mistral-7b-inst	France	2023-09	Malignant Hyperthermia	EU64.0	Low-Med
grok-4	USA	2024-12	McArdle disease (Glycogen storage disease type V)	E74.04	High
gpt-oss-120b	USA	2025-08	McArdle disease (Glycogen storage disease type V)	E74.0	Low-Med
command-r	Canada	2024-03	Exercise-induced rhabdomyolysis	M62.84	Low-Med
deepseek-chat	China	2024-12	McArdle disease (Glycogen storage disease type V)	E74.0	Medium
deepseek-r1	China	2025-01	McArdle Disease (Glycogen Storage Disease Type V)	E74.0	Medium
sonar-deep-res e	USA	2025-03	McArdle disease (Glycogen Storage Disease Type V)	E74.04	Low-Med
jamba-large-1.7	Israel	2025-07	McArdle disease (Glycogen storage disease type V)	E74.01	Low
mistral-large-2	France	2024-11	McArdle Disease (Glycogen Storage Disease Type V)	E74.01	Low-Med
command-r-plu s	Canada	2024-04	Muscular Dystrophy, Limb-Girdle Type 2A (LGMD2A or Calpainopathy)	G72.3	Low-Med
wizardlm-2-8x2 2	USA	2024-04	McArdle Disease (Glycogen Storage Disease type V)	E74.0	Low-Med
grok-2-1212	USA	2024-12	Carnitine palmitoyltransferase II deficiency	E71.310	Low-Med
gemma-2-9b-it	USA	2024-06	McArdle Disease	E70.0	Low-Med
gpt-4o	USA	2024-05	Glycogen storage disease type V (McArdle disease)	E74.0	Low-Med
llama-3.2-3b-in	USA	2024-09	McArdle disease (Glycogen storage disease type V)	E12.2	Low-Med
gpt-4o-mini	USA	2024-07	McArdle's disease (Glycogen storage disease type V)	E74.0	Low-Med
qwen-2.5-coder -	China	2024-11	Exertional Rhabdomyolysis	E76.0	Medium
claude-3-opus- 2	USA	2024-02	McArdle disease (Glycogen storage disease type V)	E74.04	Low-Med
lfm-40b	USA	2024-10	Mitochondrial myopathy	G71.0	Low-Med
gemini-2.5-pro	USA	2024-12	considerations and the key differentials		Low-Med
gemini-2.5-flas	USA	2024-12	Glycogen Storage Disease Type V (McArdle Disease)	E74.01	Low-Med

gemini-2.5-flas	USA	2024-12	McArdle Disease (Glycogen Storage Disease Type V)	E74.21	Low-Med

AI Model Bias Analysis

AI model bias analysis is generated during orchestration (Step 2). This comprehensive analysis examines cultural, geographic, and training data biases across the AI models used.

Primary Diagnosis Bias Factors:

- Cultural: Western models show stronger preference for metabolic myopathy diagnosis
- Geographic: Primarily Western diagnostic framework dominates
- Training Data: Newer models show more comprehensive differential diagnosis

Alternative Diagnoses Bias:

- Missed: Traditional Chinese Medicine patterns - Western models lack training in TCM diagnostic frameworks
- Over-diagnosed: McArdle Disease - Western models more likely to suggest this diagnosis

Bias Mitigation Recommendations:

- Cultural: Include diverse medical paradigms in model training

Critical Decision Points & Evidence Synthesis

Critical Decision Points

Key areas where models showed significant divergence in diagnostic or management approach:

Evidence Synthesis & Clinical Correlation

Symptom-Diagnosis Correlation Matrix

Symptom/Finding	McArdle Dise	Carnitine Pa
Exercise Intole	+++	+++
Muscle Weakness	+++	+++
Dark Urine Afte	+++	+++
Consanguineous	+++	+++
Exercise-Induce	++	+++

Legend: +++ Strong association, ++ Moderate, + Weak, - Not typical

Diagnostic Decision Tree

Step	Action	If Positive	If Negative
1	PYGM gene sequencing	Confirm McArdle disease diagnosis	Proceed to muscle biopsy
2	Forearm ischemic exercise test	Suggests McArdle disease	Consider other metabolic myopathies

Detailed Model Responses

Complete diagnostic assessments from each model:

1. mistral-7b-inst (France, Released: 2023-09-27)

Primary Diagnosis: Malignant Hyperthermia (ICD-10: EU64.0) - Confidence: 0.8

Reasoning: Exercise intolerance, muscle weakness, elevated CK levels, and genetic predisposition in consanguineous parents are indicative of a genetic disorder, and malignant hyperthermia is a rare genetic condi...

Differential Diagnoses:

- Hypokalemic Periodic Paralysis (ICD: E34.1) - 0.6
- Rhabdomyolysis (ICD: E37.1) - 0.5
- Metabolic Myopathies (ICD: E43) - 0.4

Key Clinical Findings:

- Exercise intolerance
- Intermittent muscle weakness
- Elevated CK levels during episodes

Recommended Tests:

- Genetic testing for malignant hyperthermia susceptibility - To confirm the diagnosis of Malignant Hyperthermia
- Electromyography (EMG) - To assess muscle function and damage
- Blood tests for electrolytes, creatinine, and myoglobin - To assess kidney function and for signs of rhabdomyolysis

Immediate Management:

- Cool the patient
- Correct electrolyte imbalances

Medications:

- Dantrolene 2.5 mg/kg IV IV
- Fluid replacement IV normal saline IV

2. grok-4 (USA, Released: 2024-12-26)

Primary Diagnosis: McArdle disease (Glycogen storage disease type V) (ICD-10: E74.04) - Confidence: 0.85

Reasoning: Episodic exercise-induced muscle weakness, pain, and dark urine (myoglobinuria) with elevated CK, in context of consanguineous parents, strongly suggest this autosomal recessive metabolic myopathy....

Differential Diagnoses:

- Carnitine palmitoyltransferase II deficiency (ICD: E71.314) - 0.6

- Tarui disease (Glycogen storage disease type VII) (ICD: E74.09) - 0.5
- Very long-chain acyl-CoA dehydrogenase deficiency (ICD: E71.310) - 0.4

Key Clinical Findings:

- Exercise-induced muscle weakness and pain with dark urine (myoglobinuria)
- Elevated CK to 5,000 during episodes, normal interictally
- Consanguineous parents suggesting autosomal recessive inheritance
- Adolescent competitive athlete with episodic symptoms

Recommended Tests:

- Serum electrolytes, BUN, creatinine, urinalysis - Assess for rhabdomyolysis complications like acute kidney injury
- Genetic testing for metabolic myopathies (e.g., PYGM gene for McArdle) - Confirm specific genetic diagnosis
- Muscle biopsy with enzyme histochemistry - Evaluate for glycogen accumulation or enzyme deficiencies

Immediate Management:

- Intravenous hydration to prevent renal complications from rhabdomyolysis
- Activity restriction: avoid intense exercise until diagnosis confirmed

Medications:

- Normal saline 1-2 L bolus followed by 200-300 mL/hr IV
- Acetaminophen 650 mg every 6 hours as needed PO

3. gpt-oss-120b (USA, Released: 2025-08-05)

4. command-r (Canada, Released: 2024-03-11)

Primary Diagnosis: Exercise-induced rhabdomyolysis (ICD-10: M62.84) - Confidence: 0.85

Reasoning: Exercise intolerance and elevated CK levels suggest exercise-induced muscle damage. Consanguinity increases risk of genetic predisposition....

Differential Diagnoses:

- McArdle's Disease (Glycogen Storage Disease Type V) (ICD: E72.1) - 0.75
- Periodic Paralysis Disorders (ICD: E83.3) - 0.6
- Mitochondrial Disease (ICD: G73.2) - 0.55

Key Clinical Findings:

- Exercise intolerance
- Elevated CK levels during episodes

Recommended Tests:

- CK levels monitoring - Confirm elevated CK and monitor response to treatment
- Muscle biopsy - Differentiate between primary muscle disorders and rhabdomyolysis
- Metabolic panel - Check for metabolic abnormalities indicative of McArdle's or mitochondrial disease

Immediate Management:

- Hydration: Encourage adequate fluid intake to prevent renal complications.
- Rest: Recommend complete physical rest until CK levels normalize.

Medications:

- Sodium bicarbonate 1-2 teaspoons (5-10mmol) in water PO

5. deepseek-chat (China, Released: 2024-12-20)

Primary Diagnosis: McArdle disease (Glycogen storage disease type V) (ICD-10: E74.0) - Confidence: 0.9

Reasoning: The patient presents with exercise intolerance, muscle weakness, and dark urine after intense exercise, which are classic symptoms of McArdle disease. The elevated CK levels during episodes further su...

Differential Diagnoses:

- Carnitine palmitoyltransferase II (CPT II) deficiency (ICD: E71.3) - 0.7
- Duchenne muscular dystrophy (ICD: G71.0) - 0.5
- Myoadenylate deaminase deficiency (ICD: E79.8) - 0.6

Key Clinical Findings:

- Elevated CK levels during episodes
- Exercise intolerance and muscle weakness

Recommended Tests:

- Genetic testing for PYGM gene mutations - Confirm diagnosis of McArdle disease
- Muscle biopsy - Assess glycogen storage and enzyme activity
- Electromyography (EMG) - Evaluate muscle function and rule out other myopathies

Immediate Management:

- Hydration to prevent renal damage from rhabdomyolysis
- Rest and avoid strenuous exercise

Medications:

- Acetaminophen 500 mg PO

6. deepseek-r1 (China, Released: 2025-01-20)

Primary Diagnosis: McArdle Disease (Glycogen Storage Disease Type V) (ICD-10: E74.0) - Confidence: 0.9

Reasoning: Exercise-induced rhabdomyolysis (dark urine, elevated CK) in a consanguineous pedigree suggests autosomal recessive metabolic myopathy. McArdle disease (GSD V) classically presents with exercise intol...

Differential Diagnoses:

- Carnitine Palmitoyltransferase II Deficiency (CPT II) (ICD: E71.3) - 0.7
- Rhabdomyolysis (Recurrent Exertional) (ICD: M62.82) - 0.4
- Mitochondrial Myopathy (ICD: G71.3) - 0.3

Key Clinical Findings:

- Consanguineous parents (first cousins)
- Exercise-triggered rhabdomyolysis (dark urine, CK 5,000)
- Episodic symptoms with normal CK between attacks

Recommended Tests:

- Genetic testing (PYGM gene for McArdle, CPT2 gene for CPT II) - Confirm specific metabolic myopathy
- Forearm exercise test with lactate/ammonia - Assess lactate response (flat curve in McArdle)
- Muscle biopsy (histochemistry/enzyme assay) - Myophosphorylase activity quantification

Immediate Management:

- Hydration during acute rhabdomyolysis
- Renal function monitoring (creatinine, urine output)

Medications:

- Acetaminophen 650-1000 mg every 6 hours PRN PO

7. sonar-deep-rese (USA, Released: 2025-03-07)

Primary Diagnosis: McArdle disease (Glycogen Storage Disease Type V) (ICD-10: E74.04) - Confidence: 0.85

Reasoning: Exercise intolerance with myalgia, myoglobinuria after intense exercise, elevated CK during episodes but normal between, autosomal recessive inheritance pattern from consanguineous parents, and typica...

Differential Diagnoses:

- Carnitine palmitoyltransferase II deficiency (ICD: E71.3) - 0.15
- Mitochondrial myopathy (ICD: G73.6) - 0.1
- Glycogen Storage Disease Type VII (Tarui disease) (ICD: E74.09) - 0.08

Key Clinical Findings:

- Exercise-induced myoglobinuria in competitive swimmer
- CK elevation during episodes but normal between episodes
- Consanguineous parents suggesting autosomal recessive inheritance
- Exercise intolerance with muscle pain triggered by high-intensity swimming

Recommended Tests:

- PYGM gene sequencing - Confirm McArdle disease diagnosis
- Plasma acylcarnitine profile - Rule out fatty acid oxidation defects like CPT II deficiency
- Forearm exercise test (non-ischemic) - Assess lactate and ammonia response to exercise

Immediate Management:

- Temporarily restrict high-intensity swimming training
- Ensure adequate hydration during any exercise
- Monitor for signs of acute kidney injury during episodes

Medications:

- Sucrose/glucose 15-30g oral PO

8. jamba-large-1.7 (Israel, Released: 2025-07-01)

Primary Diagnosis: McArdle disease (Glycogen storage disease type V) (ICD-10: E74.01) - Confidence: 0.8

Reasoning: Exercise intolerance, intermittent muscle weakness, dark urine, elevated CK levels post-exercise, and consanguinity suggest a genetic disorder involving muscle metabolism....

Differential Diagnoses:

- Duchenne muscular dystrophy (ICD: G71.0) - 0.5
- Rhabdomyolysis (ICD: M62.82) - 0.6
- Metabolic myopathy (ICD: E74.09) - 0.7

Key Clinical Findings:

- Exercise intolerance
- Intermittent muscle weakness

Recommended Tests:

- Muscle biopsy - Confirm glycogen storage disease
- Genetic testing - Confirm McArdle disease mutation
- Electromyography (EMG) - Differentiate between myopathies

Immediate Management:

- Avoid intense exercise
- Hydration with electrolytes

Medications:

- Oral glucose supplements 15g per episode PO

9. mistral-large-2 (France, Released: 2024-11-01)

Primary Diagnosis: McArdle Disease (Glycogen Storage Disease Type V) (ICD-10: E74.01) - Confidence: 0.8

Reasoning: Exercise intolerance, intermittent muscle weakness, dark urine post-exercise, elevated CK levels during episodes, and consanguineous parents suggest a genetic metabolic disorder....

Differential Diagnoses:

- Carnitine Palmitoyltransferase II Deficiency (CPT II) (ICD: E71.311) - 0.6
- Myoadenylate Deaminase Deficiency (ICD: E74.8) - 0.4
- Tarui Disease (Glycogen Storage Disease Type VII) (ICD: E74.02) - 0.3

Key Clinical Findings:

- Exercise intolerance
- Intermittent muscle weakness
- Dark urine after intense training
- Elevated CK levels during episodes

Recommended Tests:

- Genetic Testing for GSD Type V - Confirm McArdle Disease
- Muscle Biopsy - Evaluate for glycogen storage abnormalities
- Urine Myoglobin Test - Confirm myoglobinuria

Immediate Management:

- Rest and avoid intense physical activity
- Hydration and dietary modifications

Medications:

- N/A N/A N/A

10. command-r-plus (Canada, Released: 2024-04-04)

Primary Diagnosis: Muscular Dystrophy, Limb-Girdle Type 2A (LGMD2A or Calpainopathy) (ICD-10: G72.3) - Confidence: 0.75

Reasoning: The patient's symptoms of exercise intolerance, intermittent muscle weakness, and elevated CK levels during episodes are highly suggestive of a muscular dystrophy. The consanguinity of the parents and...

Differential Diagnoses:

- McArdle Disease (GSD V, Glycogen Storage Disease Type V) (ICD: E74.0) - 0.6
- Periodic Paralysis, Hypokalemic (ICD: G73.0) - 0.5
- Mitochondrial Myopathy (ICD: G73.89) - 0.4

Key Clinical Findings:

- Elevated CK levels during episodes
- Consanguineous parents
- Limited social interactions

Recommended Tests:

- Genetic Testing (CAPN3 gene analysis) - Confirm diagnosis of LGMD2A
- Muscle Biopsy - Evaluate for muscular dystrophy and metabolic myopathies
- Electromyography (EMG) and Nerve Conduction Studies (NCS) - Assess for myopathic or neuropathic processes

Immediate Management:

- Referral to a pediatric neurologist with expertise in neuromuscular disorders
- Genetic counseling for the patient and family to discuss the potential impact of consanguinity on future offspring

Medications:

- Nonsteroidal Anti-inflammatory Drugs (NSAIDs) Standard doses PO

11. wizardlm-2-8x22 (USA, Released: 2024-04-15)

Primary Diagnosis: McArdle Disease (Glycogen Storage Disease type V) (ICD-10: E74.0) - Confidence: 0.8

Reasoning: Exercise intolerance, intermittent muscle weakness, elevated CK levels, and dark urine following intense exercise are classic symptoms. The patient's parents are consanguineous, which increases the ri...

Differential Diagnoses:

- Carnitine Palmitoyltransferase Deficiency (CPT Deficiency) (ICD: E71.4) - 0.6
- Mitochondrial Myopathy (ICD: Unknown) - 0.5
- Duchenne Muscular Dystrophy (ICD: G71.0) - 0.4

Key Clinical Findings:

- Exercise intolerance with muscle weakness
- Intermittent dark urine after intense exercise
- Elevated CK levels during episodes

Recommended Tests:

- Muscle biopsy with histochemical analysis and electron microscopy - To confirm the presence of glycogen storage abnormalities
- Genetic testing for mutations in the PYGM gene - To confirm diagnosis of McArdle Disease
- Blood lactate levels during and after exercise - To evaluate for a normal or low increase in lactate, which is indicative of McArdle Disease

Immediate Management:

- Avoid strenuous exercise until diagnosis is confirmed
- Educate patient and family about symptoms and potential triggers

Medications:

- Sucrose (oral) Variable, based on patient tolerance and response PO

12. grok-2-1212 (USA, Released: 2024-12-12)

Primary Diagnosis: Carnitine palmitoyltransferase II deficiency (ICD-10: E71.310) - Confidence: 0.85

Reasoning: Exercise intolerance, muscle weakness, dark urine post-exercise, elevated CK levels, and consanguinity suggest a genetic disorder affecting fatty acid metabolism....

Differential Diagnoses:

- McArdle disease (ICD: E74.02) - 0.7
- Rhabdomyolysis due to exertional causes (ICD: M62.82) - 0.6
- Mitochondrial myopathy (ICD: G71.3) - 0.5

Key Clinical Findings:

- Exercise intolerance
- Episodes of dark urine post-exercise
- Elevated CK levels during episodes
- Consanguinity

Recommended Tests:

- Genetic testing for CPT II deficiency - Confirm diagnosis
- Muscle biopsy - Evaluate for glycogen storage disorders

- Serum acylcarnitine profile - Assess for fatty acid oxidation disorders

Immediate Management:

- Avoid strenuous exercise
- Hydrate and monitor urine output

Medications:

- Intravenous fluids Normal saline at 1.5-2 times maintenance rate IV

13. gemma-2-9b-it (USA, Released: 2024-06-27)

Primary Diagnosis: McArdle Disease (ICD-10: E70.0) - Confidence: 0.7

Reasoning: Exercise intolerance, intermittent muscle weakness, dark urine after intense training, elevated CK levels are consistent with glycogen storage disease type V (McArdle Disease). Consanguinity of paren...

Differential Diagnoses:

- Myophosphorylase Deficiency (ICD: Unknown) - 0.2
- Mitochondrial Myopathy (ICD: G35.0) - 0.1
- Rhabdomyolysis (ICD: M62.0) - 0.05

Key Clinical Findings:

- Recurrent episodes of muscle weakness and pain after exercise
- Elevated CK levels during episodes, normal between episodes
- History of consanguinity

Recommended Tests:

- Muscle biopsy - To assess for glycogen storage abnormalities and confirm McArdle Disease
- Genetic testing - To identify specific gene mutations associated with McArdle Disease

Immediate Management:

- Hydration and electrolyte replacement
- Supportive care during muscle weakness episodes

14. gpt-4o (USA, Released: 2024-05-13)

Primary Diagnosis: Glycogen storage disease type V (McArdle disease) (ICD-10: E74.0) - Confidence: 0.9

Reasoning: The symptoms of exercise intolerance, muscle weakness, episodes of dark urine, and elevated CK levels after intense physical activity are characteristic of McArdle disease, especially given the consan...

Differential Diagnoses:

- Carnitine palmitoyltransferase II deficiency (ICD: E71.3) - 0.7
- Duchenne muscular dystrophy (ICD: G71.0) - 0.4
- Myoadenylate deaminase deficiency (ICD: E79.8) - 0.3

Key Clinical Findings:

- Exercise intolerance
- Episodes of dark urine after exercise

Recommended Tests:

- Genetic testing for PYGM gene mutations - To confirm McArdle disease
- Muscle biopsy - To assess glycogen accumulation
- Acylcarnitine profile - To rule out carnitine palmitoyltransferase II deficiency

Immediate Management:

- Educate on avoiding strenuous exercise
- Hydration to prevent myoglobinuria

Medications:

- Sucrose 10-20g PO

15. llama-3.2-3b-in (USA, Released: 2024-09-25)

Primary Diagnosis: McArdle disease (Glycogen storage disease type V) (ICD-10: E12.2) - Confidence: 0.9

Reasoning: Elevated CK levels, exercise intolerance, muscle weakness, and episodes of dark urine are characteristic of McArdle disease, a rare genetic disorder affecting glycogenolysis in muscles....

Differential Diagnoses:

- Miyoshi myopathy (ICD: G93.3) - 0.3
- Hypothyroidism (ICD: E00.0) - 0.2
- Mitochondrial myopathies (ICD: G56.9) - 0.1

Key Clinical Findings:

- Elevated serum creatine kinase (CK) levels during episodes of muscle weakness
- Dark urine and ketonuria during episodes of intense exercise

Recommended Tests:

- Serum CK level - Rule out other myopathies and metabolic disorders
- Urine ketone test - Supports diagnosis of McArdle disease

Immediate Management:

- IV hydration and monitoring of electrolyte levels
- Hospitalization for nutritional support and management of symptoms

Medications:

- Mannitol 1g/kg IV bolus IV

16. gpt-4o-mini (USA, Released: 2024-07-18)

Primary Diagnosis: McArdle's disease (Glycogen storage disease type V) (ICD-10: E74.0) - Confidence: 0.85

Reasoning: The patient's exercise intolerance, muscle weakness, dark urine, and elevated CK levels after exercise are consistent with McArdle's disease, a genetic disorder of glycogen metabolism. The

consanguini...

Differential Diagnoses:

- Pompe disease (Glycogen storage disease type II) (ICD: E74.1) - 0.1
- Congenital myopathy (ICD: G72.0) - 0.05
- Hyperthyroidism (ICD: E05.9) - 0.05

Key Clinical Findings:

- Elevated CK levels to 5,000 after intense exercise
- Episodes of dark urine following strenuous activity

Recommended Tests:

- Genetic testing for glycogen storage diseases - To confirm diagnosis of McArdle's disease or other glycogen storage diseases
- Muscle biopsy - To assess for glycogen accumulation

Immediate Management:

- Educate the patient about avoiding intense exercise
- Provide hydration and dietary consultation

Medications:

- Creatine monohydrate 5 g daily PO

17. qwen-2.5-coder- (China, Released: 2024-11-12)

Primary Diagnosis: Exertional Rhabdomyolysis (ICD-10: E76.0) - Confidence: 0.8

Reasoning: Patient presents with exercise intolerance, muscle weakness, dark urine, and elevated CK levels. History of consanguinity increases risk for rare genetic conditions, but current symptoms align with ex...

Differential Diagnoses:

- Metabolic Myopathies (ICD: G71.0-G71.9) - 0.6
- Hypophosphatemic Rickets (ICD: E23.0) - 0.3
- Heat Illness (ICD: T67.0) - 0.2

Key Clinical Findings:

- Exercise intolerance and intermittent muscle weakness
- Elevated CK levels (5,000) during episodes

Recommended Tests:

- Genetic Testing - To identify genetic mutations associated with metabolic myopathies
- Electromyogram (EMG) - To assess muscle function and identify any muscle disease
- Serum and Urine Electrolytes - To rule out electrolyte imbalances

Immediate Management:

- Hydration with intravenous fluids
- Rest and avoid strenuous exercise

Medications:

- Allopurinol 100 mg PO

18. claude-3-opus-2 (USA, Released: 2024-02-29)

Primary Diagnosis: McArdle disease (Glycogen storage disease type V) (ICD-10: E74.04) - Confidence: 0.85

Reasoning: Exercise intolerance, episodic muscle weakness, myoglobinuria, and high CK levels in a young athlete with consanguineous parents suggests a glycogen storage disorder, most likely McArdle disease....

Differential Diagnoses:

- Carnitine palmitoyltransferase II deficiency (ICD: E71.318) - 0.4
- Duchenne muscular dystrophy (ICD: G71.0) - 0.3
- Mitochondrial myopathy (ICD: G71.3) - 0.2

Key Clinical Findings:

- Exercise-induced muscle pain, weakness, and myoglobinuria
- Episodic symptoms with normal strength between episodes
- Markedly elevated CK levels during acute episode
- Young athlete with consanguineous parents

Recommended Tests:

- Ischemic forearm exercise test - Assess rise in lactate and ammonia to diagnose McArdle disease
- PYGM gene sequencing - Confirm diagnosis of McArdle disease
- CK level - Monitor disease activity and risk of rhabdomyolysis

Immediate Management:

- Avoid strenuous exercise and fasting
- Ensure adequate hydration
- Provide emergency plan for acute episodes

Medications:

- Oral glucose or sucrose 37 g glucose or 75 g sucrose 5 min before exercise PO

19. lfm-40b (USA, Released: 2024-10-29)

Primary Diagnosis: Mitochondrial myopathy (ICD-10: G71.0) - Confidence: 0.8

Reasoning: The patient's symptoms are consistent with a mitochondrial myopathy, presenting with exercise intolerance, muscle weakness, and elevated CK levels, which are common in mitochondrial disorders. The con...

Differential Diagnoses:

- Duchenne muscular dystrophy (ICD: G71.0) - 0.6
- Malignant hyperthermia (ICD: T80.5) - 0.4
- Carnitine palmitoyltransferase (CPT) II deficiency (ICD: E74.4) - 0.7

Key Clinical Findings:

- Elevated CK levels during episodes

- Dark urine after intense training

Recommended Tests:

- Mitochondrial DNA analysis - Confirm mitochondrial myopathy
- Muscle biopsy - Assess muscle histology and enzyme activity
- CPT II assay - Rule out CPT II deficiency

Immediate Management:

- Rest and hydration
- Avoidance of strenuous exercise until diagnosis is confirmed

Medications:

- Coenzyme Q10 100-300 mg/day PO

20. gemini-2.5-pro (USA, Released: 2024-12-19)

21. gemini-2.5-flas (USA, Released: 2024-12-19)

Primary Diagnosis: Glycogen Storage Disease Type V (McArdle Disease) (ICD-10: E74.01) - Confidence: 0.9

Reasoning: Classic presentation with exercise intolerance, muscle pain, and dark urine (myoglobinuria) after exertion, particularly intense training. Elevated CK levels during episodes and normal between episode...

Differential Diagnoses:

- Carnitine Palmitoyltransferase II (CPT II) Deficiency (ICD: E71.321) - 0.7
- Malignant Hyperthermia Susceptibility (ICD: G71.3) - 0.4
- Duchenne Muscular Dystrophy (atypical presentation or carrier state) (ICD: G71.01) - 0.2

Key Clinical Findings:

- 16-year-old competitive swimmer with exercise intolerance
- Intermittent muscle weakness and episodes of dark urine after intense training
- CK levels elevated to 5,000 during episode, normal between episodes
- Parents are consanguineous (first cousins)

Recommended Tests:

- Forearm ischemic exercise test - To assess for failure of lactate production, characteristic of McArdle disease (due to myophosphorylase deficiency).
- Genetic testing for PYGM gene mutation - Confirmatory diagnosis for McArdle disease. Also consider CPT2 gene for CPT II deficiency.
- Urine myoglobin - To confirm rhabdomyolysis (dark urine is suggestive).

Immediate Management:

- Advise complete rest and avoidance of further strenuous exercise.
- Ensure adequate hydration to prevent renal complications from myoglobinuria.
- Monitor urine output and color; consider IV fluids if signs of acute kidney injury.

Medications:

- No specific medication for McArdle disease (management is supportive) N/A N/A

22. gemini-2.5-flas (USA, Released: 2024-12-19)

Primary Diagnosis: McArdle Disease (Glycogen Storage Disease Type V) (ICD-10: E74.21) - Confidence: 0.9

Reasoning: The patient's presentation of exercise intolerance, intermittent muscle weakness, elevated CK levels (especially after exertion), and dark urine (suggestive of myoglobinuria) in a young athlete is hig...

Differential Diagnoses:

- Mitochondrial Myopathy (ICD: G71.3) - 0.6
- Carnitine Palmitoyltransferase II (CPT II) Deficiency (ICD: E71.31) - 0.7
- Pompe Disease (Glycogen Storage Disease Type II) (ICD: E74.01) - 0.3

Key Clinical Findings:

- Exercise-induced muscle weakness and pain
- Elevated creatine kinase (CK) levels after exertion
- Dark urine after intense training (suggestive of myoglobinuria)
- Consanguineous parents

Recommended Tests:

- Muscle Biopsy with Histochemistry and Enzyme Assays - To definitively diagnose McArdle disease by demonstrating deficiency of myophosphorylase. Can also assess for other myopathies.
- Genetic Testing for PYGM gene mutations - To confirm the diagnosis of McArdle disease by identifying pathogenic mutations in the gene encoding myophosphorylase.
- Urine Myoglobin - To confirm the presence of myoglobinuria during an symptomatic episode.

Immediate Management:

- Advise patient to avoid strenuous exercise that triggers symptoms.
- Ensure adequate hydration.
- Monitor for signs of rhabdomyolysis (severe muscle pain, dark urine, decreased urine output, kidney injury).

Medications:

- No specific medication for the underlying enzyme deficiency.