

MEDLEY

Medical AI Ensemble Clinical Decision Report

Case ID: Case_4

Title: Case_4 - Medical Analysis

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Primary Diagnostic Consensus

Diagnosis	ICD-10	Agreement	Confidence	Status
McArdle disease (Glycogen storage disease type V)	E74.0	22.7%	Low	PRIMARY

Alternative & Minority Diagnoses

Diagnosis	ICD-10	Support	Type
Carnitine palmitoyltransferase II deficiency	Unknown	18.2%	Alternative (20-29%)
Duchenne muscular dystrophy	Unknown	18.2%	Alternative (20-29%)
Mitochondrial myopathy	Unknown	18.2%	Alternative (20-29%)
Rhabdomyolysis	Unknown	9.1%	Minority (<20%)
Metabolic Myopathies	Unknown	9.1%	Minority (<20%)
Malignant Hyperthermia	Unknown	4.5%	Minority (<20%)
Hypokalemic Periodic Paralysis	Unknown	4.5%	Minority (<20%)
Tarui disease (Glycogen storage disease type VII)	Unknown	4.5%	Minority (<20%)
Very long-chain acyl-CoA dehydrogenase deficiency	Unknown	4.5%	Minority (<20%)
Exercise-induced rhabdomyolysis	Unknown	4.5%	Minority (<20%)
McArdle's Disease (Glycogen Storage Disease Type V)	Unknown	4.5%	Minority (<20%)
Periodic Paralysis Disorders	Unknown	4.5%	Minority (<20%)
Mitochondrial Disease	Unknown	4.5%	Minority (<20%)
Carnitine palmitoyltransferase II (CPT II) deficiency	Unknown	4.5%	Minority (<20%)
Myoadenylate deaminase deficiency	Unknown	4.5%	Minority (<20%)
McArdle disease (Glycogen Storage Disease Type V)	Unknown	4.5%	Minority (<20%)

Diagnosis	ICD-10	Support	Type
Glycogen Storage Disease Type VII (Tarui disease)	Unknown	4.5%	Minority (<20%)
Metabolic myopathy	Unknown	4.5%	Minority (<20%)
Muscular Dystrophy, Limb-Girdle Type 2A (LGMD2A or Calpainopathy)	Unknown	4.5%	Minority (<20%)
McArdle Disease (GSD V, Glycogen Storage Disease Type V)	Unknown	4.5%	Minority (<20%)
Periodic Paralysis, Hypokalemic	Unknown	4.5%	Minority (<20%)
Mitochondrial Myopathy	Unknown	4.5%	Minority (<20%)
McArdle disease	Unknown	4.5%	Minority (<20%)
Rhabdomyolysis due to exertional causes	Unknown	4.5%	Minority (<20%)
Miyoshi myopathy	Unknown	4.5%	Minority (<20%)
Hypothyroidism	Unknown	4.5%	Minority (<20%)
Mitochondrial myopathies	Unknown	4.5%	Minority (<20%)
McArdle's disease (Glycogen storage disease type V)	Unknown	4.5%	Minority (<20%)
Pompe disease (Glycogen storage disease type II)	Unknown	4.5%	Minority (<20%)
Congenital myopathy	Unknown	4.5%	Minority (<20%)
Hyperthyroidism	Unknown	4.5%	Minority (<20%)
Exertional Rhabdomyolysis	Unknown	4.5%	Minority (<20%)
Hypophosphatemic Rickets	Unknown	4.5%	Minority (<20%)
Heat Illness	Unknown	4.5%	Minority (<20%)
Malignant hyperthermia	Unknown	4.5%	Minority (<20%)
Carnitine palmitoyltransferase (CPT) II deficiency	Unknown	4.5%	Minority (<20%)

Analysis Overview
Models Queried: 22
Successful Responses: 22
Consensus Level: Low
Total Estimated Cost: \$0.507

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	McArdle	CPT II D	Mitochon
Exercise intolerance	+++	-	-
Muscle weakness	-	-	-
Dark urine	+++	-	-
Elevated CK	-	++	-
Consanguinity	-	-	-

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

Diagnostic Decision Tree

Step	Action	If Positive	If Negative
1	PYGM gene mutation analysis	Confirm McArdle disease diagnosis	Proceed to forearm exercise test

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

- Recurrent fever episodes
- Positive family history of similar episodes

Primary Recommendations

- Consider McArdle disease (Glycogen storage disease type V) among differential diagnoses
- Avoid strenuous exercise
- Ensure adequate hydration
- Obtain Genetic testing for PYGM gene mutations for diagnostic confirmation

Primary Diagnosis Clinical Summaries

Orchestrated analysis not available for this case.

Diagnostic Landscape Analysis

Detailed Diagnostic Analysis

The ensemble analysis identified **McArdle disease (Glycogen storage disease type V)** as the primary diagnosis with 22.7% consensus among 0 models.

Detailed Alternative Analysis

Diagnosis	Support	Key Evidence	Clinical Significance
Carnitine palmitoyltransferase II deficiency	18.2%	0 models	Less likely
Duchenne muscular dystrophy	18.2%	0 models	Less likely
Mitochondrial myopathy	18.2%	0 models	Less likely
Rhabdomyolysis	9.1%	0 models	Unlikely
Metabolic Myopathies	9.1%	0 models	Unlikely
Malignant Hyperthermia	4.5%	0 models	Unlikely
Hypokalemic Periodic Paralysis	4.5%	0 models	Unlikely
Tarui disease (Glycogen storage disease type VII)	4.5%	0 models	Unlikely

Minority Opinions

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

- **Rhabdomyolysis** (ICD-10: Unknown) - 9.1% agreement (0 models)

Supporting Models:

- **Metabolic Myopathies** (ICD-10: Unknown) - 9.1% agreement (0 models)

Supporting Models:

- **Malignant Hyperthermia** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Hypokalemic Periodic Paralysis** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Tarui disease (Glycogen storage disease type VII)** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Very long-chain acyl-CoA dehydrogenase deficiency** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Exercise-induced rhabdomyolysis** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **McArdle's Disease (Glycogen Storage Disease Type V)** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Periodic Paralysis Disorders** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Mitochondrial Disease** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Carnitine palmitoyltransferase II (CPT II) deficiency** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Myoadenylate deaminase deficiency** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **McArdle disease (Glycogen Storage Disease Type V)** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Glycogen Storage Disease Type VII (Tarui disease)** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Metabolic myopathy** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Muscular Dystrophy, Limb-Girdle Type 2A (LGMD2A or Calpainopathy)** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **McArdle Disease (GSD V, Glycogen Storage Disease Type V)** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Periodic Paralysis, Hypokalemic** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Mitochondrial Myopathy** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **McArdle disease** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Rhabdomyolysis due to exertional causes** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Miyoshi myopathy** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Hypothyroidism** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

• **Mitochondrial myopathies** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **McArdle's disease (Glycogen storage disease type V)** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Pompe disease (Glycogen storage disease type II)** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Congenital myopathy** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Hyperthyroidism** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Exertional Rhabdomyolysis** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Hypophosphatemic Rickets** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Heat Illness** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Malignant hyperthermia** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

- **Carnitine palmitoyltransferase (CPT) II deficiency** (ICD-10: Unknown) - 4.5% agreement (0 models)

Supporting Models:

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
Forearm ischemic exercise test	Assess lactate response	Routine	As indicated

Treatment Recommendations

Treatment recommendations pending diagnostic confirmation.

Model Diversity & Bias Analysis

Model Response Overview & Cost Analysis

Model	Origin	Tier	Cost	Diagnosis	Training Profile
mistral-7b-inst	France	Budget	<\$0.01	Malignant Hyperthermia	General
grok-4	USA	Premium	\$0.048	McArdle disease (Glycogen storage disease type V)	Alternative
gpt-oss-120b	USA	Mid-Range	<\$0.01	McArdle disease (Glycogen storage disease type V)	Standard
command-r	Canada	Mid-Range	<\$0.01	Exercise-induced rhabdomyolysis	Standard
deepseek-chat	China	Budget	<\$0.01	McArdle disease (Glycogen storage disease type V)	Regional
gemini-2.5-pro	USA	Premium	\$0.033	considerations and the key differentials	General
deepseek-r1	China	Budget	<\$0.01	McArdle Disease (Glycogen Storage Disease Type V)	Regional
sonar-deep-rese	USA	Premium	\$0.028	McArdle disease (Glycogen Storage Disease Type V)	Standard
jamba-large-1.7	Israel	Premium	\$0.023	McArdle disease (Glycogen storage disease type V)	Standard
gemini-2.5-flas	USA	Budget	<\$0.01	McArdle Disease (Glycogen Storage Disease Type V)	General
mistral-large-2	France	Premium	\$0.029	McArdle Disease (Glycogen Storage Disease Type V)	Standard
command-r-plus	Canada	Premium	\$0.041	Muscular Dystrophy, Limb-Girdle Type 2A (LGMD2A or Calpainopathy)	Standard
wizardlm-2-8x22	USA	Mid-Range	<\$0.01	McArdle Disease (Glycogen Storage Disease type V)	Standard
grok-2-1212	USA	Premium	\$0.027	Carnitine palmitoyltransferase II deficiency	Standard
gemma-2-9b-it	USA	Budget	<\$0.01	McArdle Disease	Standard
gpt-4o	USA	Premium	\$0.048	Glycogen storage disease type V (McArdle disease)	Comprehensive
gemini-2.5-flas	USA	Budget	<\$0.01	Glycogen Storage Disease Type V (McArdle Disease)	General
llama-3.2-3b-in	USA	Budget	<\$0.01	McArdle disease (Glycogen storage disease type V)	General
gpt-4o-mini	USA	Budget	<\$0.01	McArdle's disease (Glycogen storage disease type V)	General
qwen-2.5-coder-	China	Mid-Range	<\$0.01	Exertional Rhabdomyolysis	Regional
claude-3-opus-2	USA	Premium	\$0.202	McArdle disease (Glycogen storage disease type V)	Comprehensive
lfm-40b	USA	Mid-Range	<\$0.01	Mitochondrial myopathy	Standard

****Total Estimated Cost: \$0.507****

Understanding Training Profiles

Training profiles indicate the type and depth of medical knowledge in each model:

Comprehensive: Extensive medical literature training with broad clinical knowledge

Standard: Standard medical knowledge base with general clinical training

Regional: Region-specific medical training reflecting local practices and conditions

General: Broad general knowledge, not specifically trained on medical literature

Alternative: Alternative medical perspectives and non-conventional approaches

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 tendency to diagnose rare genetic disorders
- Geographic: Western approach dominates due to model distribution
- Training Data: Newer models show more nuanced understanding of metabolic disorders

Alternative Diagnoses Bias:

- Missed: Traditional Chinese Medicine patterns - Western models lack training in TCM diagnostic approaches
- Over-diagnosed: McArdle Disease - Western models more likely to suggest rare genetic disorders

Bias Mitigation Recommendations:

- Geographic: Include more diverse medical perspectives

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 condition.

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

7. 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 intolerance...

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

8. 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

9. 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

10. 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.

11. 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

12. 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

13. 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

14. 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

15. 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

16. 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

17. 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

18. 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

19. 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

20. 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

21. 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

22. Ifm-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