Together with Mito we can Take on TK2d

Thymidine kinase 2 deficiency (TK2d):
A rare, life-threatening, genetic mitochondrial disease (mito) characterized by progressive and severe muscle weakness (myopathy), which can impact the ability to walk, eat, and breathe independently.[1,2,3]

OVERVIEW OF MITOCHONDRIAL DISEASE

What Are Mitochondrial Diseases?

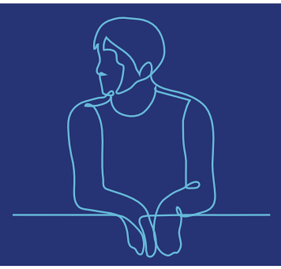
Mitochondrial diseases are a group of rare, life-threatening, genetic conditions that affect the parts of our body that need the most energy – muscles, the heart, and the brain.[4,5]

3 or more organ systems not functioning properly is a hallmark characteristic of mitochondrial disease.[4]

It is estimated that

1 in 5,000

people have some form of mitochondrial disease.[6]



What Causes Mitochondrial Diseases?

Mitochondria are found in nearly every cell of the body. They generate the energy needed for proper functioning of organs.[4]

Mitochondria may malfunction when the genes encoding them, found in either the mitochondria or nucleus, undergo harmful changes.[7,8]

These changes in genes are called mutations, and they may be inherited or occur spontaneously (acquired) during a person's lifetime.[7,8]

> **Disability puts you in situations you would not otherwise experience, with people you would never meet, and with abilities you would never discover.**
>
> – Parent
>
> *of school-aged child with TK2d*

Patient advocacy organizations and community pages can provide valuable information, helpful resources, and support for those affected by TK2d.

learn more

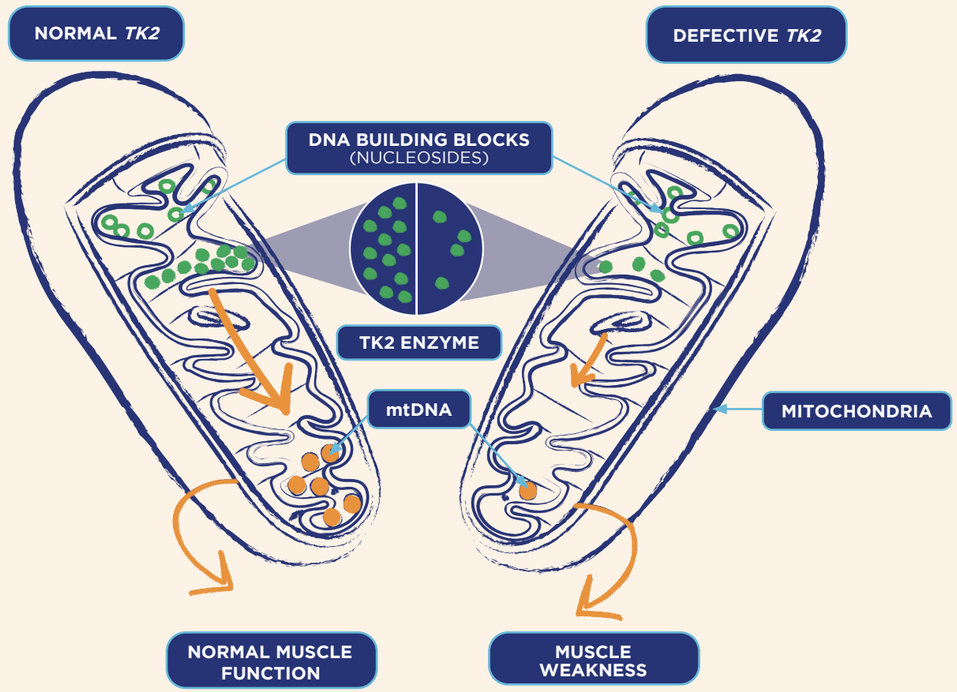
TK2d.com

OVERVIEW OF TK2d

TK2d: A Rare Mitochondrial Disease

Thymidine kinase 2 deficiency (TK2d) is a rare primary mitochondrial myopathy, meaning it presents predominantly as progressive and severe muscle weakness (myopathy) and low muscle tone (hypotonia).[1,2,3]

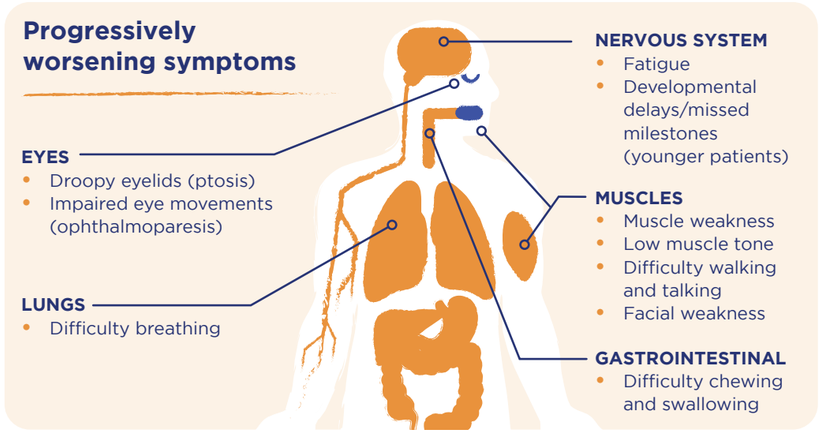
- More specifically, TK2d is caused by an error (mutation) in the *TK2* (thymidine kinase 2) nuclear gene and is inherited in an autosomal recessive manner, meaning both parents must pass along the mutated *TK2* gene.[1,3]
- However, not all children whose parents are carriers of the mutated *TK2* gene will develop TK2d; each child has a 25% chance of having TK2d.[9]



***TK2* gene mutations reduce the amount and quality of mitochondrial DNA (mtDNA) within cells,[10] leading to insufficient energy production and improper function of muscles and organs.**[4]

SIGNS, SYMPTOMS, AND COURSE OF THE DISEASE

TK2d can present in different ways and affect different parts of the body.[1,2,3]



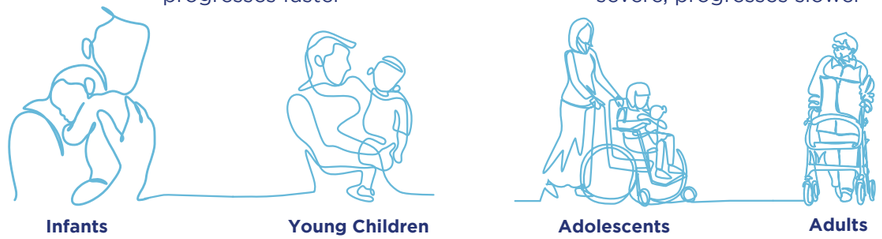
Symptoms can differ from person to person and can present at any age from infancy up until late adulthood.[1]

Patients with TK2d typically experience progressive proximal muscle weakness, which is a root cause of most symptoms, including the loss of motor skills and respiratory difficulties.[9,10,11] Eventually, patients may lose the ability to walk, eat, and breathe independently.[1,2,3]

Symptoms can progress slowly or quickly, depending on each person and age of onset.

Early onset: often more severe, progresses faster

Later onset: often less severe, progresses slower



> **I am afraid these things will eventually lead to me losing my window into the world. I already can't speak. What if I could no longer communicate via my laptop?**
>
> —Adult
> *with TK2d*

Although these symptoms may seem overwhelming, **they can be managed with support** from a dedicated healthcare team, practical strategies, and a variety of resources.

DIAGNOSIS

TK2d diagnosis can be challenging due to variable symptoms that overlap with similar diseases, such as muscular dystrophy, Pompe disease, spinal muscular atrophy (SMA), and others.[1,5,10]

The above signs could be an indication of TK2d. It is recommended to seek advice from a healthcare professional about testing to confirm a suspected diagnosis of TK2d.[1,12]

> **I first started having symptoms around puberty. I started out with a liver specialist, who sent me to a neurologist, who sent me to a geneticist. They did a muscle biopsy and saw ragged red fibers. Then they did a blood test and found the TK2d gene. It took about 3 years to get the diagnosis.**
>
> —Young Adult
> *with TK2d*

Genetic testing is the most direct path to diagnosing TK2d.[1]

Only certain genetic tests can definitively confirm a diagnosis of TK2d.[9]

For suspected TK2d, different genetic testing options are available, such as whole-genome sequencing, whole-exome sequencing, single-gene testing, and multigene panels that include the *TK2* gene.[9]

Other tests are often ordered for patients who show symptoms of TK2d, including:[9,10]

- **Blood tests**
- **Muscle biopsy**
- **Brain magnetic resonance imaging (MRI)**
- **Electromyography (EMG) test**

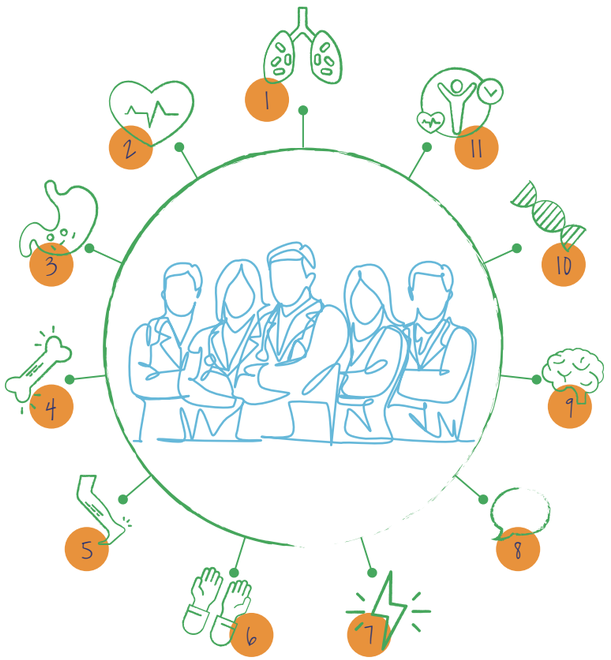
An early, accurate diagnosis of TK2d is important to inform best supportive care and potential involvement in clinical trials.[1,2,3]

The TK2d community is small but mighty, forming strong and supportive networks through close connections with individuals diagnosed around the world.

UNDERSTANDING THE MULTIDISCIPLINARY HEALTHCARE TEAM

Currently, TK2d management is centered around supportive care.[6]

The team will work together to manage the symptoms and optimize the quality of life for those living with TK2d.[9,12,13,14]



1. **Pulmonologists and respiratory therapists**
Assist with breathing difficulties.

2. **Cardiologists**
Assess and manage heart muscle weakness.

3. **Gastroenterologists and nutritionists**
Manage feeding difficulties, assist in dietary guidance and address special food needs.

4. **Orthopedic specialists**
Help with muscle weakness, bone abnormalities or joint problems.

5. **Physical therapists**
Focus on adjusting and adapting movement, strength, and coordination.

6. **Occupational therapists**
Focus on improving the ability to perform activities of daily living.

7. **Metabolic specialists**
Monitor and manage energy levels.

8. **Speech therapists**
Help with speaking difficulties and strengthening the facial muscles required to chew.

9. **Neurologists**
Help with challenges related to movement.

10. **Clinical geneticists**
Provide more information into the cause of health challenges.

11. **Pediatricians or primary care physicians**
Provide regular health and wellness checkups and diagnose and treat a wide range of general health conditions.

learn more

To learn more about ongoing clinical trials for mitochondrial disease, visit:

UMDF Clinical Trials Webpage

ClinicalTrials.gov

MANAGING TK2d

To help manage TK2d symptoms and address muscle weakness, a healthcare team may recommend **medical equipment and devices**, such as:[9,14]

> **With TK2d, it is not a sprint, it is more like a marathon. So, you take everything a little bit at a time. People say that practice makes perfect. With TK2d, practice makes progress.**
>
> – Parent
> *of a toddler with TK2d*

- **Adaptive eating utensils and other aids for feeding**
- **Back braces**
- **Breathing support devices, such as passive ventilators, continuous positive airway pressure (CPAP) and bilevel positive airway pressure (BiPAP) machines**
- **Communication devices (e.g., text to speech, hearing aids)**
- **Feeding tubes, including gastrostomy tubes (G-Tube), gastrojejunostomy (GJ) tubes, and nasogastric (NG) tubes**
- **Leg immobilizers**
- **Supramalleolar orthoses (SMOs), ankle foot orthoses (AFOs), and other orthotic solutions**
- **Visual support tools**
- **Wheelchairs/walkers**

> **Something as simple as putting on your shirt by yourself, being able to reach the light switch or roll over in bed at night to get comfortable instead of relying on someone else. I long to just feel like I'm capable of doing stuff again.**
>
> – Young Adult
> *with TK2d*

Patient Community Organizations and Resources play a critical role in supporting and connecting individuals with similar experiences, as well as providing information and educational resources.[5]

United Mitochondrial Disease Foundation (UMDF)
UMDF can connect individuals with a variety of helpful resources, educational information, and strong, supportive shoulders to lean on.

WEBSITE

support@umdf.org
(888) 900-6486
@theUMDF
@UMDF

The Jeremiah Gracen TK2d Foundation
The Jeremiah Gracen TK2d Foundation is committed to spreading hope for those affected by TK2d by educating, raising awareness, and being a liaison for support services.

WEBSITE

"Tell Me, Teach Me, Is it TK2D?" written by The Jeremiah Gracen TK2d Foundation.
@JeremiahGracen
TK2DWarrior

MitoAction
MitoAction is striving to make a measurable impact in the lives of those affected by mitochondrial diseases through support, education, and advocacy.

WEBSITE

mito411@mitoaction.org
(888) 648-6228
MyMito App
@mitoaction

Muscular Dystrophy Association (MDA)
MDA provides access to support and programs that empower the lives of people living with neuromuscular diseases through 150+ multi-disciplinary MDA Care Centers, community education programs, camp and recreation, gene therapy support, advocacy opportunities, and more.

WEBSITE

ResourceCenter@mdausa.org
1-833-ASK-MDA1 (1-833-275-6321)

Mitochondrial Medicine Society (MMS)
MMS is an international group of physicians, researchers, and clinicians working towards advancing education, research, and global collaboration in clinical mitochondrial medicine.

WEBSITE

This brochure was created in partnership with

ucb Inspired by patients. Driven by science.
United Mitochondrial Disease Foundation
The Jeremiah Gracen TK2D Foundation

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