Rare Disease Donor Guide

Primary Human Skeletal Muscle Cells – Donors with Neuromuscular & Other Rare Disorders

COOK® MYOSITE

The Right Cells for Your Research

Cook MyoSite is proud to provide dependable, highly characterized primary human skeletal muscle cells to researchers around the world. Powered by expertise developed from our clinical research, our cells are known for their consistent and reliable performance in a wide range of research applications.

Cook MyoSite offers a wide range of cells isolated from a diverse spectrum of donors, including donors affected with neuromuscular and other rare disorders. In this guide, you can learn more about these disorders and browse our current cell inventory specific to each disorder.

We understand the value that accurate representation provides to your research. If you're looking for a cell donor affected by a disorder that is not included here, we want to hear from you! Our experienced donor and tissue procurement team is ready to work with you and your unique needs.

Ready to purchase now? Scan the QR code to the right or email us at researchsales@cookmyosite.com.

- Myotonic Dystrophy
- Duchenne Muscular Dystrophy
- Chronic Fatigue Syndrome
- Myasthenia Gravis
- Multiple Sclerosis
- Amyotrophic Lateral Sclerosis
- Lambert-Eaton (LEMS)
- Guillain-Barre Syndrome
- Marfan Syndrome
- Friedrich's Ataxia

Myotonic Dystrophy (DM)

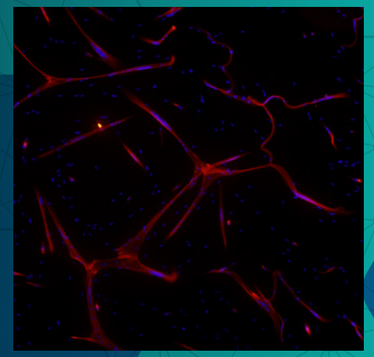
DM is a hereditary disease that causes progressive muscle weakness and is the most common adult-onset muscular dystrophy. There are two types, DM1 and DM2.

Genetic Info & Symptoms:

- DM1 is caused by the amplification of a triplet CTG repeat in the DMPK gene on Chromosome 19. Normal repeat is 5-37. Affected individuals have >50 repeats.
- DM2 is caused by a CCTG tetranucleotide repeat in the ZNF9 gene. Affected individuals have >75 repeats.

Cell Culture Considerations:

- Mostly normal growth and differentiation, reduced differentiation capacity in some donors



Myosin Heavy Chain (red), DAPI (blue)

Available Samples

AGE	SEX	ETHNICITY	CATALOG NUMBER	TISSUE OF ORIGIN	DIABETES	BMI	TOBACCO USE
18	F	Caucasian	SK-2222-P03023-18F	Vastus Lateralis	N	20	N
19	M	Caucasian	SK-2222-P03025-19M	Vastus Lateralis	N	28	N
50	F	Caucasian	SK-2222-P03022-50F	Vastus Lateralis	N	20	Y
62	M	African American	SK-2222-P03007-62M	Vastus Lateralis	Y	27	N

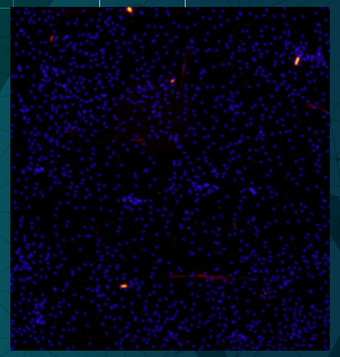
*Additional donors and detailed medical/genetic information available in our inventory

Duchenne Muscular Dystrophy (DMD)

DMD is a genetic disease characterized by agressive muscle degeneration and caused by defects in the dystrophin-producing DMD gene. DMD almost always affects males and presents at a young age.

Genetic Info & Symptoms:
- Numerous different types of of mutations in the dystophin gene have been identified (>1000) including point mutations, deletions, and insertions. - Rapid muscle degeneration.

Cell Culture Considerations:
- Cells were obtained from individuals in the late stage of disease whose muscle tissue was almost depleted of myogenic cells
- Cells are almost devoid of the myogenic marker Desmin and exhibit poor differentiation and MHC expression.



Myosin Heavy Chain (red), DAPI (blue)

Available Samples

AGE	SEX	ETHNICITY	CATALOG NUMBER	TISSUE OF ORIGIN	DIABETES	BMI	TOBACCO USE
20	M	Caucasian	SK-2222-P01294-20M	Vastus Lateralis	N	26	N
21	M	Caucasian	SK-2222-P01543-21M	Vastus lateralis	N	48	N

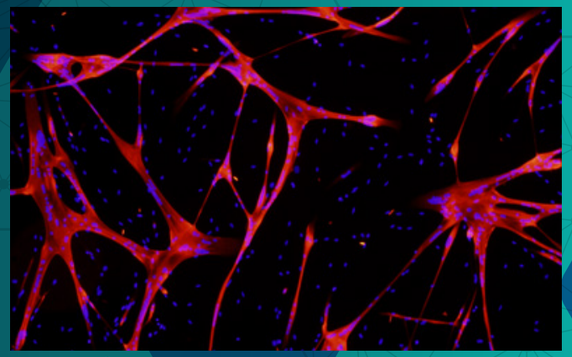
*Additional medical information available in inventory

Chronic Fatigue Syndrome (CFS)

CFS is a poorly understood disease characterized by extreme fatigue that does not improve with sleep or rest. While many theories exist, the cause of CFS remains unknown.

Genetic Info & Symptoms:
- No known genetic cause
- Extreme fatigue that does not improve with rest

Cell Culture Considerations:
- N/A



Myosin Heavy Chain (red), DAPI (blue)

Available Samples

AGE	SEX	ETHNICITY	CATALOG NUMBER	TISSUE OF ORIGIN	DIABETES	BMI	TOBACCO USE
24	M	Caucasian	T02398-24M	Vastus lateralis	Unknown	20	Unknown
39	F	Asian	T02421-39F	Vastus lateralis	Unknown	24	Unknown
49	F	Caucasian	T02390-49F	Vastus lateralis	Unknown	25	Unknown
59	M	Caucasian	T02400-59M	Vastus lateralis	Unknown	N/A	Unknown

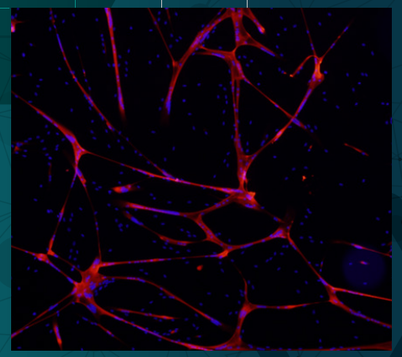
*Additional donors and medical information available in our inventory

Myasthenia Gravis (MG)

MG is an autoimmune disease in which antibodies alter acetylcholine receptors at the neuromuscular junctions. This interrupts normal muscle contraction and causes the symptoms of this disease.

Genetic Info & Symptoms:
- This does not appear to be a directly inheritable condition, though there are associated haplotypes with early onset MG
- Dysphagia, dysarthria, dyspnia, diplopia, ptosis, ocular myasthenia

Cell Culture Considerations:
- N/A



Myosin Heavy Chain (red), DAPI (blue)

Available Samples

AGE	SEX	ETHNICITY	CATALOG NUMBER	TISSUE OF ORIGIN	DIABETES	BMI	TOBACCO USE
22	M	African American	SK-2222-P03019-22M	Vastus Lateralis	N	28	Unknown
50	F	African American	SK-2222-P03020-50F	Vastus Lateralis	N	28	N
52	F	African American	SK-2222-P03010-52F	Vastus Lateralis	Y	27	N
56	F	Caucasian	SK-2222-P03014-56F	Vastus Lateralis	N	31	N

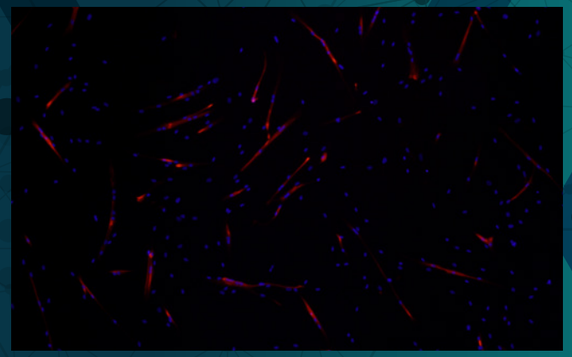
*Additional medical information available in our inventory

Multiple Sclerosis (MS)

MS is a central nervous system and autoimmune disease in which the body's immune system attacks its own nerves' myelin sheath.

Genetic Info & Symptoms:
- No known genetic cause
- Symptoms vary widely but include vision problems, loss of muscle function, fatigue

Cell Culture Considerations:
- N/A



Myosin Heavy Chain (red), DAPI (blue)

Available Samples

AGE	SEX	ETHNICITY	CATALOG NUMBER	TISSUE OF ORIGIN	DIABETES	BMI	TOBACCO USE
67	F	Caucasian	SK-2222-P01315-67F	Vastus Lateralis	N	16	N

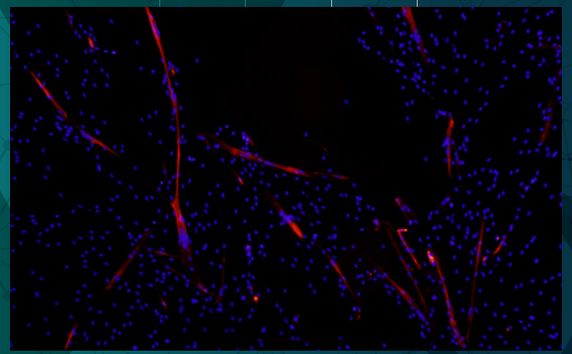
*Additional medical information available in our inventory

Amyotrophic Lateral Sclerosis (ALS)

ALS, also known as Lou Gehrig's disease, is a progressive neurological disease that results in the death of motor neurons in the brain and spinal cord.

Genetic Info & Symptoms:
- ~90% of cases are sporadic; the remaining 10% can be linked to genetic mutations
- Dysphagia, dysarthria, dyspnea, muscle dysfunction

Cell Culture Considerations:
- N/A



Myosin Heavy Chain (red), DAPI (blue)

Available Samples

AGE	SEX	ETHNICITY	CATALOG NUMBER	TISSUE OF ORIGIN	DIABETES	BMI	TOBACCO USE
62	M	Caucasian	T01623-62M	Vastus Lateralis	N	21	Y

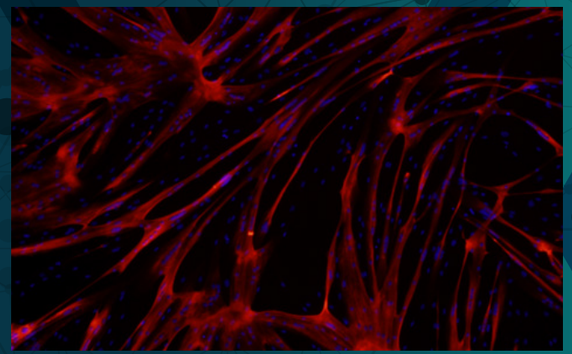
*Additional medical information available in our inventory

Lambert-Eaton Myasthenic Syndrome (LEMS)

LEMS is an autoimmune disease in which the body's immune system attacks its own tissues at the neuromuscular junction, interrupting normal communication between the nervous and muscular systems.

Genetic Info & Symptoms:
- Roughly half of cases linked to cancer; the other half have an unclear cause
- Loss of some calcium channels at nerve endings

Cell Culture Considerations:
- N/A



Myosin Heavy Chain (red), DAPI (blue)

Available Samples

AGE	SEX	ETHNICITY	CATALOG NUMBER	TISSUE OF ORIGIN	DIABETES	BMI	TOBACCO USE
59	F	Caucasian	SK-2222-P03008-59F	Vastus Lateralis	N	25	Y

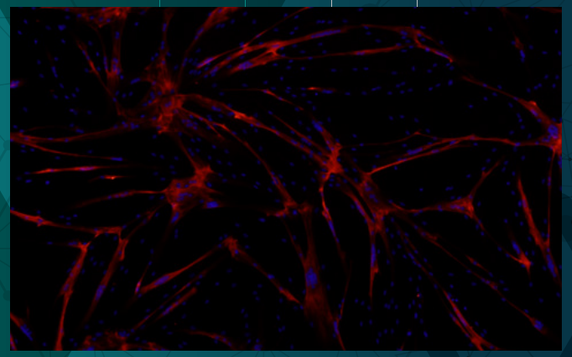
*Additional medical information available in our inventory

Guillain-Barre Syndrome (GBS)

GBS is an autoimmune disease in which the body's immune system attacks its nervous system. GBS can be acute at the onset, and most patients recover significant motor function after 6 months.

Genetic Info & Symptoms:
- No known genetic cause
- Damage to myelin sheath resulting in inability to walk, loss of muscle function, double vision

Cell Culture Considerations:
- N/A



Myosin Heavy Chain (red), DAPI (blue)

Available Samples

AGE	SEX	ETHNICITY	CATALOG NUMBER	TISSUE OF ORIGIN	DIABETES	BMI	TOBACCO USE
57	F	Caucasian	SK-2222-P01255-57F	Rectus Abdominis	N	48	N

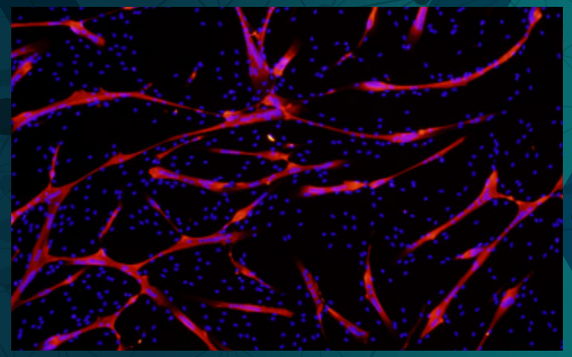
*Additional medical information available in our inventory

Marfan Syndrome

Marfan Syndrome is a genetic disease that affects connective tissues throughout the body. Symptoms of Marfan Syndrome vary widely but frequently include vision problems and aortic aneurysms.

Genetic Info & Symptoms:
- Mutations in FBN1 gene leading to reduced fibrillin-1
- Decrease in functional microfibrils and connective tissue elasticity

Cell Culture Considerations:
- N/A



Myosin Heavy Chain (red), DAPI (blue)

Available Samples

AGE	SEX	ETHNICITY	CATALOG NUMBER	TISSUE OF ORIGIN	DIABETES	BMI	TOBACCO USE
38	M	Caucasian	SK-2222-P01434-38M	Vastus lateralis	N	16	Y

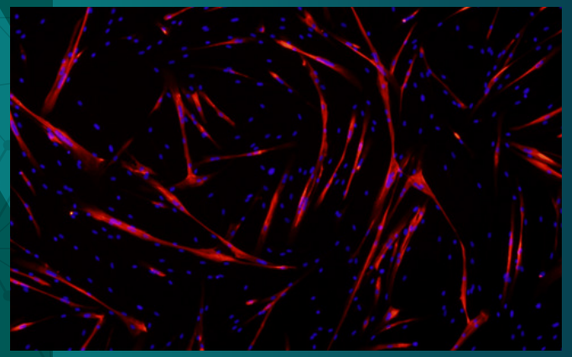
*Additional medical information available in our inventory

Friedrich's Ataxia (FRDA)

FRDA is a genetic disease that affects the nervous system. FRDA is caused by mutations in the FXN gene leading to lower levels of frataxin in certain nerve and muscle cells. This is believed to interrupt normal mitochondrial functions and cause the symptoms of FRDA.

Genetic Info & Symptoms:
- GAA trinucleotide repeat expansion in FXN gene
- Progressive ataxia, sensory loss, dysarthria, cardiomyopathy

Cell Culture Considerations:
- N/A



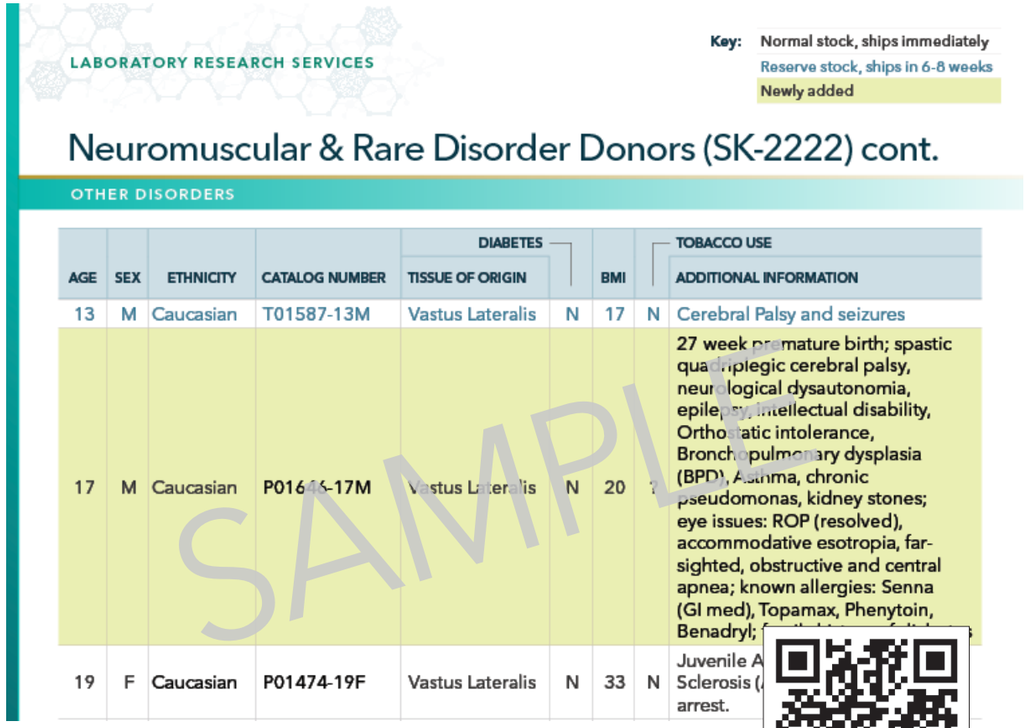
Myosin Heavy Chain (red), DAPI (blue)

Available Samples

AGE	SEX	ETHNICITY	CATALOG NUMBER	TISSUE OF ORIGIN	DIABETES	BMI	TOBACCO USE
49	M	Caucasian	SK-2222-P01423-49M	Vastus lateralis	Y	49	Y

*Additional medical information available in our inventory

See the full inventory for detailed information



LABORATORY RESEARCH SERVICES

Key: Normal stock, ships immediately
Reserve stock, ships in 6-8 weeks
Newly added

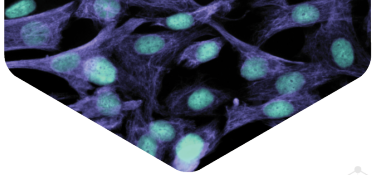
Neuromuscular & Rare Disorder Donors (SK-2222) cont.

OTHER DISORDERS

AGE	SEX	ETHNICITY	CATALOG NUMBER	DIABETES: TISSUE OF ORIGIN		BMI	TOBACCO USE	ADDITIONAL INFORMATION
13	M	Caucasian	T01587-13M	Vastus Lateralis	N	17	N	Cerebral Palsy and seizures
17	M	Caucasian	P01646-17M	Vastus Lateralis	N	20		27 week premature birth; spastic quadriplegic cerebral palsy, neurological dysautonomia, epilepsy, intellectual disability, Orthostatic intolerance, Bronchopulmonary dysplasia (BPD), Asthma, chronic pseudomonas, kidney stones; eye issues: ROP (resolved), accommodative esotropia, far-sighted, obstructive and central apnea; known allergies: Senna (GI med), Topamax, Phenytoin, Benadryl;
19	F	Caucasian	P01474-19F	Vastus Lateralis	N	33	N	Juvenile A Sclerosis (arrest.

SAMPLE

Scan the QR code or visit
https://www.cookmyosite.com/skmdc-skeletal-muscle-cells-dl



Making regenerative medicine a part of everyday medicine.