VALOSIN-CONTAINING PROTEIN (VCP) ASSOCIATED MULTISYSTEM PROTEINOPATHY (MSP)

PREVIOUSLY KNOWN AS: INCLUSION BODY MYOPATHY WITH EARLY-ONSET PAGET DISEASE AND FRONTOTEMPORAL DEMENTIA (IBMPFD)

ABOUT THE DISEASE FOR PATIENTS AND FAMILIES

CURE VCP DISEASE, INC.

FEATURES

This genetic disease can affect the muscles, bones, nerves, and brain. Individuals with this condition typically do not develop symptoms until mid-adulthood and may only exhibit one symptom. Symptoms vary from person to person, even among family members. It is unknown how many people are affected with this condition in the world, but it is extremely rare. Even though disease-modifying therapies do not exist for many of the conditions, interventions and supportive therapies can help improve quality of life.

Multi-disciplinary care is vital to screen and treat the various symptoms that may develop over a person's lifetime. Work with your team of doctors and therapists to develop an individualized, comprehensive care plan.

INHERITANCE

This condition is inherited in an autosomal dominant pattern, which means that an affected individual has a 50% chance of passing the VCP mutation along to a child.

MULTI-DISIPLINARY TEAM

The care team may include: (depending on your conditions)

- Neurologist
- Endocrinologist
- Psychologist
- Pulmonologist
- Cardiologist
- Geneticist
- General Practitioner
- Physical Therapist
- Occupational Therapist
- Speech Language Pathologist
- Respiratory Therapist
- Genetic Counselor
- Social Worker
- Caregiver/Family Support

POTENTIAL SYMPTOMS

Tell your medical provider if you are experiencing

These symptoms:

- Pain
- Weakness
- Muscle loss
- Muscle cramps, spasms, twitches, or tremors
- Tingling in hands or feet
- Recent falls
- Bone fractures or deformities
- Hearing problems
- Trouble swallowing
- Shortness of breath
- Heart problems
- Fatigue

Difficulty with:

- Walking or climbing stairs
- Standing up
- Lifting and carrying heavy things
- Housework and yardwork
- Handwriting or typing
- Toileting and hygiene
- Frequent urgency to use the bathroom
- Eating
- Sleeping
- Communication
- Engaging in social activities
- Change in behavior or mood

DIAGNOSIS

A mutation in the VCP gene causes multisystem proteinopathy 1 (MSP1). Genetic testing remains the only definitive way to diagnose this condition. The VCP protein has a wide variety of functions within cells, and a variety of conditions may occur in an individual when a VCP gene variant is present.

CARE GUIDELINES

Scan for publication in the Orphanet journal of rare diseases

JOIN THE PATIENT REGISTRY

Your participation helps advance therapeutic development.
www.curevcp.patient-registry

CONNECT WITH OTHER FAMILIES

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CLINICAL MANIFESTATIONS OF VALOSIN-CONTAINING PROTEIN (VCP) ASSOCIATED MULTISYSTEM PROTEINOPATHY (MSP)

OMIM #167320

ABOUT THE DISEASE FOR DOCTORS AND PROVIDERS

CURE VCP DISEASE, INC.

Phenotype	System affected	Clinical features	Prevalence
Inclusion body myopathy	Muscle	Axial and proximal weakness progressing distally is most common, although presentations resembling facioscapulohumeral muscular dystrophy, oculopharyngeal muscular dystrophy, and distal myopathy have been described.	~90%
Paget disease of bone	Skeletal	Bone pain, bone deformities, pathological fractures, hearing loss.	~40%
Frontotemporal dementia (FTD)	Cognitive	Rapidly progressive behavioral impairment, executive dysfunction, language impairment. Often associated with Parkinsonian features such as dystonia, tremor, gait disturbance.	~30%
Respiratory dysfunction	Pulmonary	Recurrent respiratory infections, weak cough, aspiration, sleep disordered breathing, respiratory failure due to advanced myopathy or ALS.	40-50%
Amyotrophic lateral sclerosis (ALS)	Upper and lower motor neurons	Multifocal weakness, hyperreflexia and/or areflexia, atrophy, fasciculations, bulbar weakness, respiratory muscle involvement, weight loss.	~10%
Parkinson disease	Central nervous system	Hypokinetic movement disorder, autonomic dysfunction, various non-motor features.	4%
Alzheimer disease	Cognitive	Dementia with predominant amnestic and higher order cognitive dysfunction.	2%
Spastic paraplegia	Upper motor neurons	Length-dependent weakness, hyperreflexia, spasticity, clonus.	Isolated cases
Charcot Marie Tooth disease (CMT2Y)	Peripheral nerves	Length-dependent weakness, muscle atrophy and sensory loss. Trophic foot changes and distal areflexia.	Isolated cases
Cardiomyopathy	Cardiac	Exertional shortness of breath, heart failure.	Uncertain
Dysphagia and dysarthria	Bulbar dysfunction	Impaired swallowing function, reduced speech volume, and intelligibility due to advanced myopathy or ALS.	Uncertain
Urinary and anal incontinence	Genitourinary, gastrointestinal	Urinary incontinence, anal incontinence or dysfunction	Uncertain

Table A: Korb, Manisha et al. “Development of a standard of care for patients with valosin-containing protein associated multisystem proteinopathy.” Orphanet journal of rare diseases vol. 17,1 23. 29 Jan. 2022, doi:10.1186/s13023-022-02172-5