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

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

ABOUT THE DISEASE FOR PATIENTS AND FAMILIES

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-DISCIPLINARY TEAM

The care team may include:

- 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

1302 Watson Blvd #1015
Warner Robins, GA 31093 USA
info@curevcp.org
www.curevcp.org
@curevcpdisease
## Clinical Manifestations of Valosin-Containing Protein (VCP) Associated Multisystem Proteinopathy (MSP)

**OMIM #167320**

### About the Disease for Doctors and Providers

Axial and proximal weakness progressing distally is most common, although presentations resembling facioscapulohumeral muscular dystrophy, oculopharyngeal muscular dystrophy, and distal myopathy have been described.

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