

Sporadic Inclusion Body Myositis (sIBM): An Information Sheet for General Practitioners (03/05/2015)

Introduction: Sporadic inclusion body myositis (sIBM) is a muscle disease – a type of muscular dystrophy. Most frequently acquired myopathy seen after age 50. A poorly understood, relentlessly progressive disorder. Weakness and atrophy of both proximal and distal skeletal muscles develop over months or years, leading to profound disability (wheelchair use). No effective treatment exists.

Incidence: sIBM is age-related. Typical onset is ~50-60 with wide variation: 20% show symptoms before 50. Prevalence is ~5-15/million in the population, rising to ~50+/million in those over 50.

Presentation: Age of onset varies from the early forties on. Presentation varies widely. People are affected in slightly differing ways, to different degrees, and at various rates. Progression rate varies in different muscle groups, and the overall rate of progression also varies widely. Progression tends to be more rapid in men and in those with late onset disease. The quadriceps are often affected first; toe drop, falling, and tripping are common first symptoms. For some, sIBM begins with weakness in the wrists and fingers, causing difficulty pinching, buttoning, making a fist, and gripping objects. Commonly affected muscles: finger flexors, quadriceps, lower leg muscles with relative sparing of shoulder and hip abductors and neck muscles. Does not affect heart muscle. Muscle pain is seldom noted in the literature but commonly reported by patients. Commonly seen more often in men.

Differential diagnosis: sIBM is very commonly initially misdiagnosed as polymyositis: if prednisone is administered with no improvement, further investigation is indicated. sIBM weakness comes on over months or years and progresses steadily (polymyositis tends to have an onset of weeks or months). Other forms of muscular dystrophy (e.g. limb girdle, Becker) must be considered as well.

Diagnosis: CK levels are often raised (at most ~10X normal). EMG studies usually display abnormalities. Muscle biopsy may show several abnormalities, e.g.; inflammatory cells invading cells, vacuolar degeneration, and inclusions or plaques of abnormal proteins. A blood test for antibodies against cN1A (or NT5C1A) is now available. Even with a biopsy, sIBM can be difficult to diagnose.

Treatment: No standard course of treatment has been shown to be effective or even helpful. sIBM is resistant to immunosuppressive treatment. Corticosteroids, cytotoxic-immunosuppressive agents, anti-TNF agents, interferon beta, and intravenous gamma globulin (IVIg), have all been tried.

Management: Management is symptomatic with early awareness and monitoring of potential complications (depression, fatigue, dysphagia and respiratory involvement). Prevention of falls is an early, vital consideration. Integration of palliative care may be helpful at end stages.

Complications: Cause of death commonly relates to respiratory dysfunction, aspiration, dysphagia, or cachexia. In up to 85% of cases, patients develop progressive, often severe dysphagia. Respiratory dysfunction caused by diaphragmatic weakness may present as sleep apnea: reduced lung volumes may raise carbon dioxide levels in arterial blood (hypoventilation causes hypercapnia).

Pathology: sIBM muscle displays 2 major parallel problems: 1) an autoimmune process (muscle fibers express MHC-I antigens triggering invasion by CD8+ lymphocytes) 2) a degenerative process with accumulation of many (some 29) pathological proteins within the muscle cell.

Causes: No cause or initial trigger event is known. Major theories: inflammation-immune reaction or degenerative protein disorder. Complex interplay of environment, genetic susceptibility and aging is implied. Sporadic IBM is not seen as a genetic disorder but may be predisposed by genetic factors.

References: Please see <http://www.ibmmyositis.com> for further information and references.

Written by Bill Tillier, reviewed by Dr. Steven Greenberg, Harvard University Medical School, Boston.