

# IBM Clinical Features

Population & Pathology				
Type	Incidence	Age at Onset	Age at Death	Symptoms
<b>s-IBM</b> (myocitis)	~20/10 <sup>6</sup>	> 55		Proximal muscle weakness, Dysphagia > 50% cases, Primary endomysial inflammation
h m e y r o e p d a i t t h a y r . y .	<b>IBM1</b> Desminopathy	~15/10 <sup>6</sup>	> 20	Skeletal Muscle Weakness, Arrhythmias, Heart failure
	<b>IBM2</b>	~200/10 <sup>6</sup>	~20-40	Quadriceps & Triceps Sparing
	<b>IBM3</b>	~1/10 <sup>9</sup>	~30-50	Cong Joint Contractures resolves Ext. Ophthalmoplegia Type 2A (MyHC-IIA)
	<b>IBMPFD</b>	~100/10 <sup>6</sup>	~25	~60-80

# IBM Genetics

Type	Genetics	Location	Etiology	
s-IBM	Polygenic & Multifactorial, HLA DR3, or genomic region between DR – C4		?Autoimmune Super antigen Complex	
h e r e d i t a r y	IBM1 Desminopathy	Autosomal Dominant, Some recessive reported	2q35 (desmin), 11q22 (CRYAB)	Myofibrillary myopathy associated with desmin accumulation
	IBM2	Autosomal Recessive GNE, loss of function mutation	9p12-p11	?Glycosylation defect Unconfirmed
	IBM3	Autosomal Dominant MyHC-IIa	17p13.1	Structural Mechanical Defect
	IBMPFD	Autosomal Dominant Valosin Containing Protein	9p13-p12 (p97)	?Agro some Pathway Unconfirmed

# IBM References

Type	OMIM # <a href="http://www.ncbi.nlm.nih.gov">http://www.ncbi.nlm.nih.gov</a>	CPT&ICD9 Codes	Web Site
s-IBM	147421		
h e r e d i t a r y	IBM1 Desminopathy	601419	
	IBM2	600737, 605820, 603824, 600737	<a href="http://www.hibm.org/arm/home">www.hibm.org/arm/home</a>
	IBM3	605637, 160740	
	IBMPFD	601023, 167320	1012 728.2.x or 335.x?