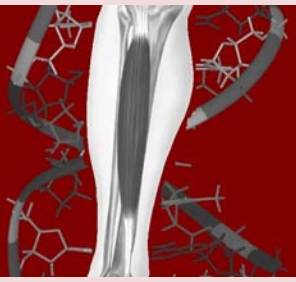

Bethlem - Myopathie

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Through this website I would like to give courage to all those who are affected and those who are still searching. Perhaps unresolved questions will be answered, new ideas put into practice and maybe even friendly new contacts made with others who are affected.

What is Bethlem myopathy? (referred to hereafter as BM)

BM is a rare genetic progressive muscular disorder with an autosomal dominant pattern of inheritance caused by a variation in type VI collagen (mutations in the COL6A1, COL6A2 or COL6A3 genes). That means every carrier of altered genetic information is affected. For each child carrying this information, the probability of being affected is around 50%. BM occurs at any age. It is often already evident in the newborn, for others it occurs much later. The main emphasis is on rapid muscle fatigue.

Important criteria exist in the contractures (restricted movements) of the fingers, wrists, elbows and ankles. Some of those affected have skin abnormalities and wounds take longer to heal. Internal organs, such as the heart and lungs, are not affected, although there are exceptions. Globally, there are at present less than 100 people known to be affected.