



Bethlem - Myopathie

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After the doctor has established the essential characteristics of Bethlem myopathy, there are two options to confirm the diagnosis.

- Variation in IV collage in one of the three genes (COL6A1, COL6A3 or COL6A3) can be determined unambiguously and *with certainty* via a blood test in a genetics laboratory.
- The alternative is a muscle biopsy.

Medical imaging techniques (X-ray, CT, MRI) and EMG (electromyography) do not deliver a clear result.