

Atypical Correlation between CTG Repeat Size Variation and Clinical Manifestation in Brothers of Myotonic Dystrophy

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The copy numbers of the CTG repeats are known to relate to the severity of clinical symptoms for myotonic dystrophy. The positive correlation between clinical manifestations and CTG repeat size has been demonstrated previously. A genetically confirmed myotonic dystrophy patient with 90 CTG repeat number had more severe clinical manifestation than brother with 120 CTG repeats, in adulthood.

Key Words: CTG repeat, Myotonic dystrophy

1. (Anticipation)
2. CTG 19q13.2-q13.3 25 가 (Muscular Disability Rating Scale: MDRS grade 4; moderate to severe proximal muscle weakness)
3. 50 3000 가 가
4. CTG 가 (High plantar arch) 5
5. 가 가 2
6. CTG 가 100 71% (Myotonic discharge)
7. 가 CTG
8. CTG 가

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G3, G4, G4+, 가

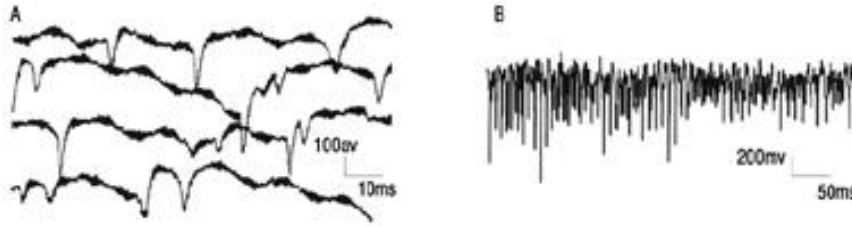


Figure 1. His EMG findings were indicative of myopathy and myotonic discharge. (A) Positive sharp wave. (B) Myotonic discharge is a spontaneous discharge of a muscle fiber that waxes and wanes in both amplitude and frequency.

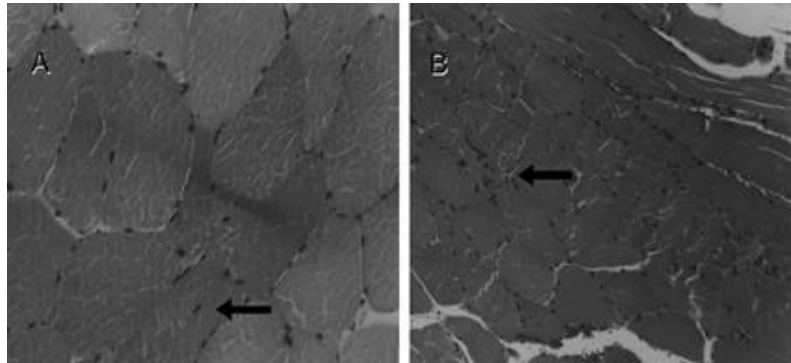


Figure 2. Right rectus femoris muscle biopsy finding. (A) You can find a internal nucleus (arrow). (B) It shows muscle size variations and floccular change (arrow).

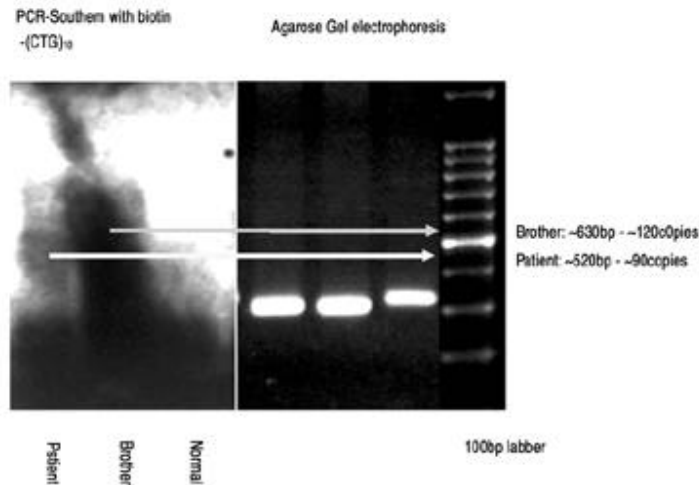


Figure 3. Southern blot analysis of (CTG)_n using serum. CTG copy number of patient is 90 copies and one of his brother is 120 copies.

1+ , , Grade 4+
 , ,
 (Tibialis anterior muscle) (Pollicis brevis muscle)
 (Myotonic discharge) (Figure 1).
 3
 Hematoxylin -Eosin (H&E) Modified Gomori trichrome (A few internal nuclei) (Floccular change) 가
 (Figure 2).
 19 CTG Steinert (Varient) Thomsen Batten .
 8

(Glucose tolerance)

Fascioscapulohumoral
가
19q13.2-13.3
Linkage analysis 가⁹
3 CTG
가¹⁰
(Internal
nucleus) (Chain forma-
tion of internal nuclei), Sarcoplasmic mass
Ring fibers, 가 Endomysial,
Interfascicular fibrous tissue 가,
1
Dive-bomber sound
Myotonic discharge Low amplitude, Short-
duration, Polyphasic motor unit potentials,
Positive sharp waves, Fibrillation potentials
Myotonic discharge
Positive sharp wave가 Dive-bomber sound

CTG 가 CTG 가⁴
CTG CTG

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