Maastricht University

Myotonic dystrophy type 1

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PROPOSITIONS

MYOTONIC DYSTROPHY TYPE 1

CLINICAL GENETICS AND MULTISYSTEM INVOLVEMENT

Isis B.T. Joosten

1. Male carriers of myotonic dystrophy type 1 (DM1) pre- and protomutations have a higher risk of symptomatic offspring compared with female small-sized repeat expansion carriers. *This thesis.*
2. Routine ECG and 24 h Holter monitoring play an essential role in the screening for cardiac involvement in DM1, and can be applied to determine the need for an invasive electrophysiological study**.** *This thesis.*
3. Screening for multiorgan involvement should be performed independent of DM1 subtype. *This thesis.*
4. Total energy expenditure in DM1 is mostly reduced due to a sedentary lifestyle, rather than to inherent metabolic abnormalities. *This thesis.*
5. Adequate follow-up of patients affected by a chronic disease requires an organized, proactive, dedicated, multi-disciplinary, and patient-centered healthcare environment.
6. Until a definite gene-based therapy becomes available, early recognition and symptomatic treatment remain the cornerstone of care for DM1-affected individuals.
7. Timely treatment of multisystemic involvement will transform DM1 from a lifespan-limiting disease to a chronic disorder.
8. Individuals affected by a rare disease face two battles. One being the illness itself, and the other, living in a world where so few people understand what you are up against. *Anonymous.*
9. Toughness is in soul and spirit, not in muscles*. Alex Karras.*
10. Great minds do not think alike. They challenge each other to think again. *Adam Grant.*