MYOPATHIES AND HEART INVOLVEMENT

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MRC Centre for Neuromuscular Diseases

NHS National Institute for Health Research

Francesco Muntoni: disclosures

Duchenne

- CI of 4 antisense oligonucleotides clinical trials with Sarepta.
- PI of Sarepta Phase III AAVrh74 micro-dystrophin
- CI of Genethon AAV8 micro-dystrophin

Spinal Muscular Atrophy

- PI of Ionis/ Biogen antisense oligonucleotide nusinersen Phase III study (Shine)
- PI of Avexis AAV9 phase III trial in SMA 1 (STRIVE-EU)-
  PI of Avexis AAV9 phase III trial in presymptomatic SMA 1-2 (SPRINT)
- PI of Roche risdiplam phase III trial in SMA 2 and 3 (Jewelfish)

Myotubular Myopathy

- PI of upcoming AAV8 MTM1 gene replacement trial

- **Other financial disclosures**
- Member of Pfizer rare disease SAB
- SAB participation for Avexis/Novartis, Biogen, Dynacure, Dyne Therapeutics, Roche, Sarepta

Cardiac involvement in myopathic conditions

Dilated cardiomyopathy

Cardiac conduction system defects

Hypertrophic cardiomyopathy

Restrictive cardiomyopathy

Cardiac involvement in myopathic conditions

- Role of different class of proteins on risk to develop specific types of cardiomyopathy
- Importance of precise diagnosis for individualised monitoring and management plans, and implementation of standards of care
- Multidisciplinary involvement of
  - neurologist / pediatrician
  - cardiologist
  - respiratory physician
  - palliative care physician
- - anaesthetist
- Role of experimental therapies under development

References

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McNally EM, Mestroni L. Dilated Cardiomyopathy: Genetic Determinants and Mechanisms. Circ Res. 2017 Sep 15;121(7):731-748.

van der Bijl P Risk Stratification of Genetic, Dilated Cardiomyopathies Associated With Neuromuscular Disorders: Role of Cardiac Imaging..Circulation. 2018 Jun 5;137(23):2514-2527.