

Additional file 2.pdf – Review of multisystemic symptoms described in literature.

| Gene/ Symptom | DES | CRYAB | FLNC | MYOT | ZASP | FHL-1 | BAG3 | DNAJB6 | TTN |
|--------------------------------|--|---|--|-------------|-------------------------------|--------------------------|----------------------|---------------|-------------------------------------|
| Cardiac involvement | AF: Arias et al. 2006 Arrhythmias: [34, 41] Conduction defects: [8, 27, 37, 39, 40] Cao et al. 2012 MI/TI: Dalakas et al. 2000 HCM: [33] DCM: [20] RCM: [32] Vrabie et al. 2005 Arbustini et al. 2006 ARVC: [38] HTx: Klauke et al. 2010 SCD: [22, 29, 35] | [9, 31, 47, 61] Iganaki et al. 2006 van der Smagt et al. 2014 | [30, 42, 60] Tasca et al. 2012 Avila-Smirnow et al. 2010 | [4, 10] | [12, 49] Vatta et al. 2003 | Binder et al. 2012 | [14, 36, 46, 48, 58] | [15] | [26] |
| Respiratory involvement | [8, 20, 27, 32, 33, 34, 35, 37, 39, 40, 44] Dagvadorj et al. 2003 Dalakas et al. 2000 | [19, 28, 31] Del Bigio et al. 2011 | [11, 30, 60] Tasca et al. 2012 | [4] | | Feldkirchner et al. 2013 | [14, 36, 46, 48, 58] | | [16, 26, 45] Edström et al. 1990 |
| Poly-neuropathy | | | [11] | [10] | [12] | [13] | [14, 36, 46, 48] | | |
| Dysphagia | [8, 20, 22, 27, 29, 33, 37, 39, 40, 43, 44] Dagvadorj et al. 2003 | [9, 28, 31, 47] | | | | | | | [45] |
| Dysarthria | [22, 27, 33] | [31] | Tasca et al. 2012 | [4] | | | [14] | | [45] |
| Cataract | [22, 33] | [9, 31] van der Smagt et al. 2014 | | | | | | | |
| Diarrhoe | [37, 39, 40, 41] | | [42, 60] | | | | | | |
| Scleroderma | | [28] | | | | | | | |
| Hearing impairment | [39, 40] | | | | [49] | | | | |

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|------------------------------|----------|------|-------------------|--|--|--------------------------|--|--|--|
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| Gynaeko- mastia | [29, 37] | [47] | | | | | | | |
| Lipo- dystrophy | | | | | | Feldkirchner et al. 2013 | | | |
| Cerebellar ataxia | | | Tasca et al. 2012 | | | | | | |

AF, atrial fibrillation; TI/MI, tricuspid or mitral insufficiency; HCM, hypertrophic cardiomyopathy; DCM, dilated cardiomyopathy; RCM, restrictive cardiomyopathy; ARVC, arrhythmogenic right ventricular cardiomyopathy; HTx, heart transplantation; SCD, sudden cardiac death, for abbreviations of genes see text.

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