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Comorbidities and mortality of persons with adult-onset myotonic dystrophy (DM1) – a Danish register-based study (Study I)

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Aim

To provide knowledge on time of diagnosis, comorbidities, and mortality in a national cohort of persons diagnosed with myotonic dystrophy (DM1) in adulthood, identified in national health registers.

Conclusion

The mean age at diagnosis for the adult-onset DM1 diagnosis was 43 years.

Individuals with adult-onset DM1 had more comorbidities than the general population. Although survival for individuals with DM1 have improved in the last 30 years in Denmark, mean age of death was 58 years in the study population.

The burden of illness due to the burden of comorbidities and the excess hazard of death calls for attention from health professionals to improve rehabilitation and survival for this population.

Background

Adult-onset myotonic dystrophy (DM1) is characterised by a diagnostic delay due to milder physical symptoms than the infantile and juvenile forms. Despite a milder phenotype of adult-onset DM1, there is a risk of negative biopsychosocial consequences particularly due to the cognitive impact. Consequently, persons with DM1 may receive less attention and have lower adherence to vital hospital follow-ups, increasing the risk of adverse events or early death.

Methods and materials

Data were extracted from the Danish National health and administrative registers in the period 1994-2022. Information on migration and death were obtained from the Migration Register and the Cause of Death Register. Data on healthcare utilisation were obtained from the Danish National Patient Register (NPR). Each person with DM1 was matched with ten reference individuals from the general Danish population with no DM1 diagnosis.

Results

Analyses were based on a population of 949 adult persons with DM1 (476 F, 473 M) and 9427 controls (fig 1).

Median age at time of diagnosis was 43 years. 40 % of individuals had a parent-children relationship; 80/161 parents were diagnosed with DM1 prior to their children; 81/161 were diagnosed after one of their children had been diagnosed. Respiratory insufficiency and cataracts were the most prevalent comorbidities in this population with DM1, with a prevalence of 32% and 30%, respectively (Table 1).

311 persons with DM1 died during the study period with an average age at death of 58 years. The mortality hazard was 5.87 higher than controls (p<0.001). Persons with DM1 and cardiovascular disease had a higher mortality rate compared to their reference groups (p<0.001) (fig. 2). Although respiratory insufficiency was more than ten times as prevalent in the DM1 population compared to controls, there was no difference in mortality rates between the two groups (p>0.9). We observed a reduction in the hazard of mortality for persons with DM1 in later years of the study period (fig. 3).

Figure 1: Flowchart of inclusion/exclusion of study population

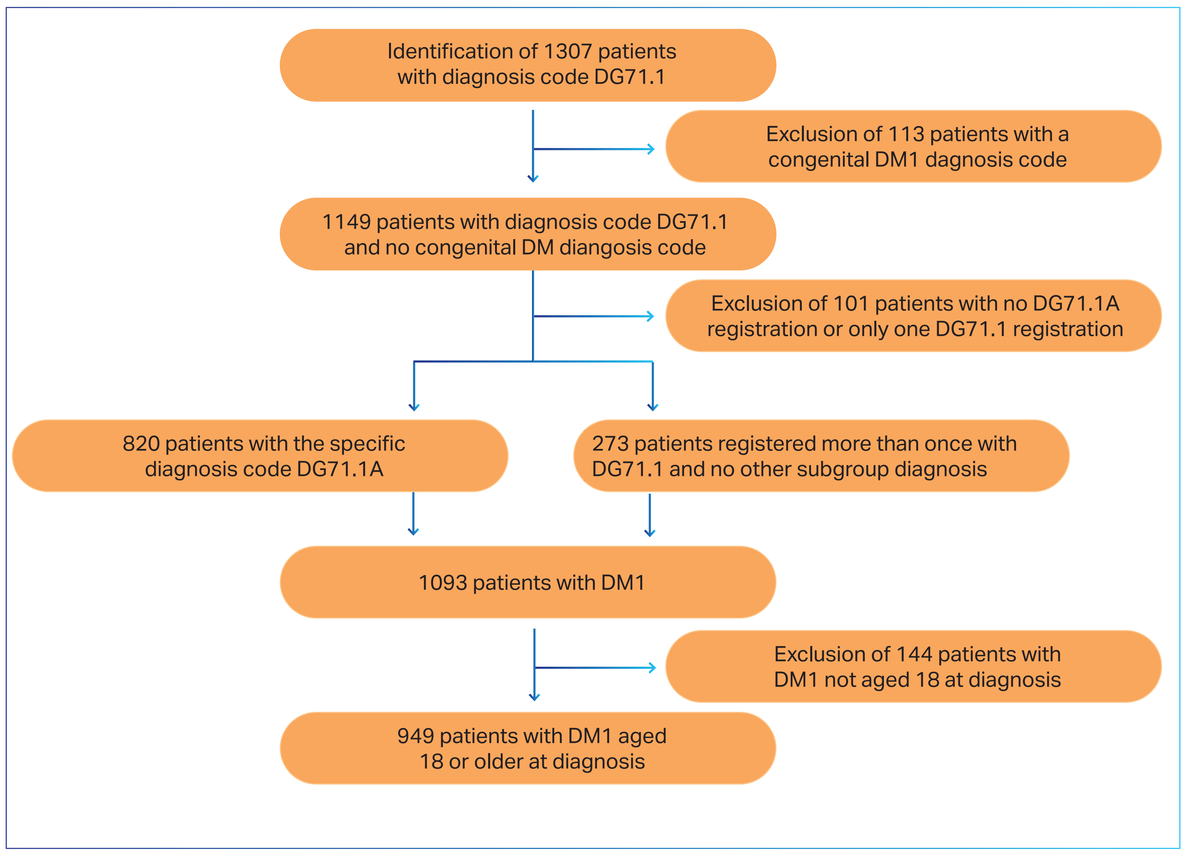

Figure 2: Overall survival among persons with DM1 and cardiovascular disease (CVD) and their controls

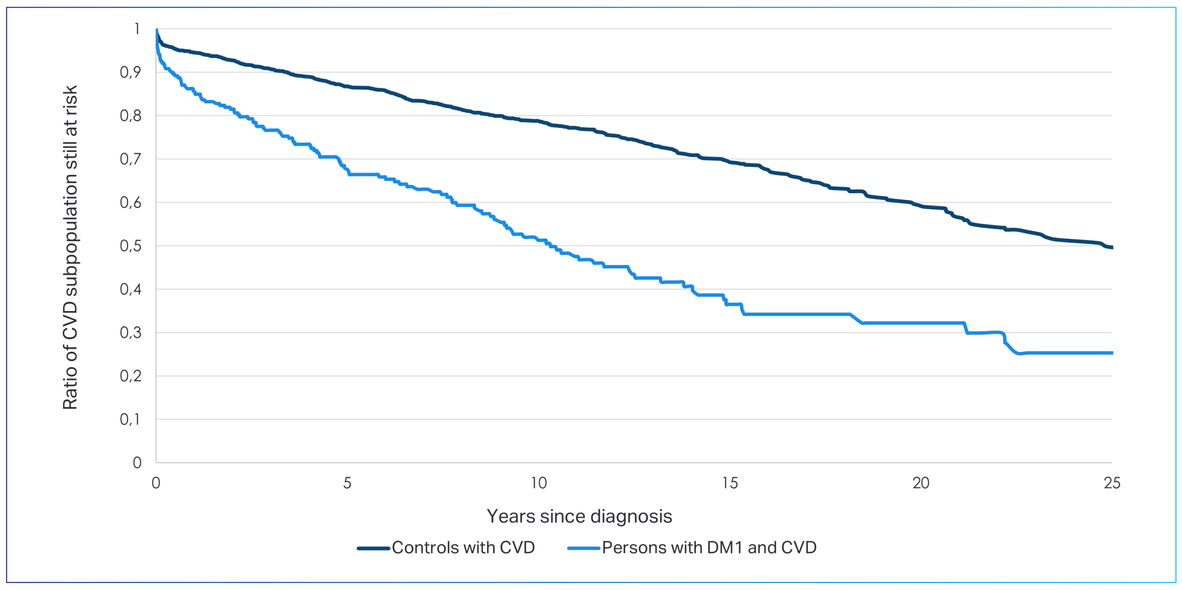

Figure 3: Risk of death in persons with DM1 stratified by year of diagnosis

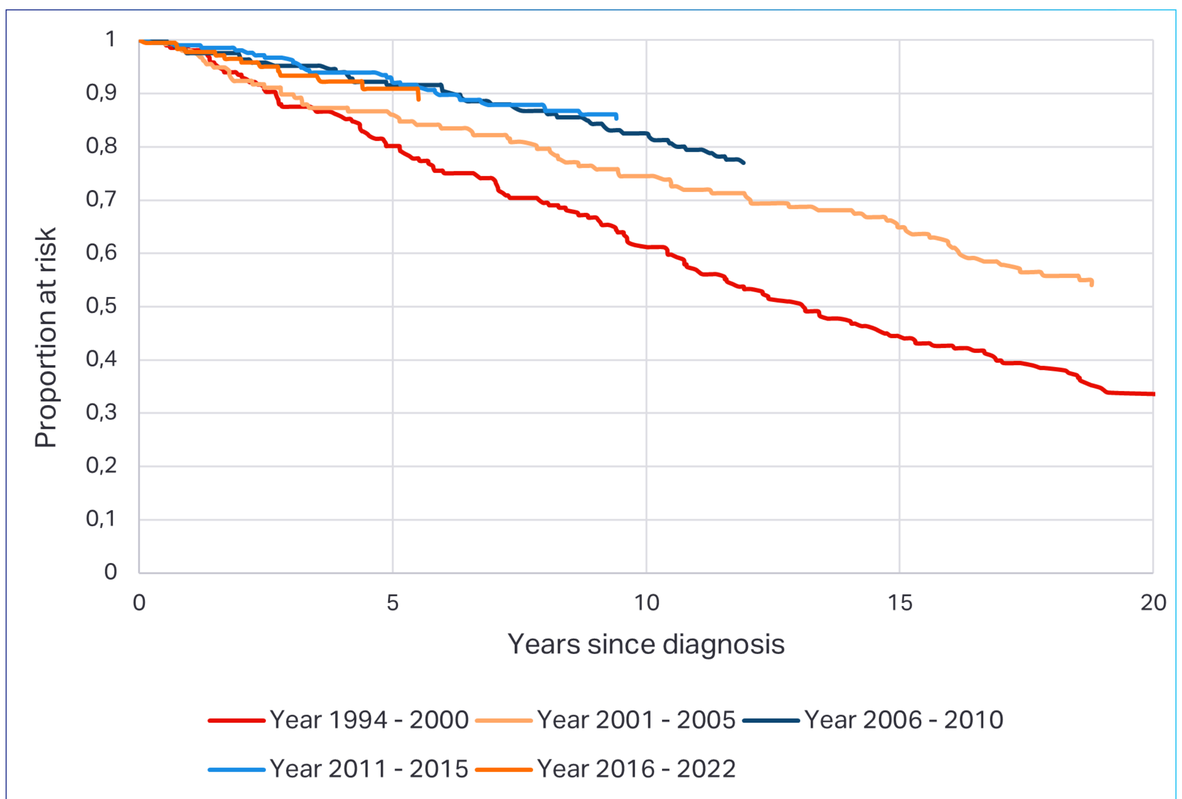

Table 1: The ten most frequent comorbidities observed for persons with DM1 and their controls

Diagnosis	Persons with DM1, n = 948	Controls, n = 9,396	P-value
Respiratory insufficiency	302 (32%)	111 (1.2%)	<0.001
Cataracts (age-related and other kinds)	282 (30%)	480 (5.1%)	<0.001
Pain in abdomen and pelvis	153 (16%)	988 (11%)	<0.001
Knee, shin and ankle fracture	106 (11%)	316 (3.4%)	<0.001
Atrial fibrillation and auricular flutter	100 (11%)	288 (3.1%)	<0.001
Eyelid disorders	87 (9.2%)	108 (1.1%)	<0.001
Cardiac insufficiency	65 (6.9%)	178 (1.9%)	<0.001
Type 2 diabetes	59 (6.2%)	242 (2.6%)	<0.001
Non-specifc pain	44 (4.6%)	447 (4.8%)	0.9
Female infertility	34 (3.6%)	185 (2.0%)	<0.001

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