Egen classification revisited in SMA

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The EK scale was developed as a clinical tool to assess functional ability and to determine the need for, and impact of intervention in nonambulatory duchenne muscular dystrophy (DMD) and spinal muscular atrophy (SMA). EK is a composite scale consisting of 10 items that are clinically relevant to the non-ambulatory patients. Each item is divided into four grades, scored 0–3 and the sum of scores of the items is the overall functional level. In reliability, validity and sensitivity studies published 1995–2002 the EK scale was shown to (1) discriminate between individuals, (2) predict the need of assisted ventilation in DMD, (3) detect change of function as a result of (a) the natural history of the two diseases or (b) scoliosis surgery and spinal bracing. However, EK was not as sensitive to change over time in individuals with SMA as in individuals with DMD.

Advances in drug treatments mean there is greater need for robust and sensitive tools that measure life events. The purpose of the present study was to (1) improve the discriminatory power of EK in SMA and (2) increase the number of clinically meaningful items. An international group of 10 experts identified 10 new items representing aspects of disease progression characteristic for the non-ambulatory stages of SMA. After evaluating these new items in 58 patients from Denmark, Italy and the UK they were reduced to seven and the manual revised. The seven new items were tested for discriminatory and content validity in conjunction with the original scale in 81 non-ambulatory patients from the three countries, aged 2–70 years. Preliminary tests show that the sum of scores of EK was significantly correlated with both FVC% ($rs = 0.77$, $p < 0.01$) and muscle strength measured as MMT ($rs = 0.87$, $p < 0.01$). Further results from this international, multi-centre study supported by treat NMD will be presented.

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