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Cure Myotonic Dystrophy UK: Facilitating and Accelerating Clinical Research in Congenital Myotonic Dystrophy

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Cure DM
UK Myotonic Dystrophy Patient Registry
University College London Hospitals NHS Foundation Trust
UCL

Disclosures

I, NIKOLETTA NIKOLENKO have no financial relationships to disclose concerning the content of this presentation or session.

Background

- Myotonic Dystrophy type 1 (DM1) is the most common adult muscular dystrophy affecting 8,000 people in the UK.
- Congenital myotonic dystrophy (CDM) is the most severe form of myotonic dystrophy type 1 (DM1) usually manifesting at birth.
- CDM similar to DM1, is an autosomal dominant neuromuscular disorder that is characterised by multisystem involvement and progressive nature.
- Advances in the understanding of the molecular pathogenesis of DM1 have enabled the development of a potential new targeted treatment for patients with CDM.

Objective: To study the demographic and clinical characteristics of CDM in UK.

Methods

- CureDM is a UK registered charity that aims to support patients and families affected by congenital myotonic dystrophy type 1 (CDM) and raise awareness for this condition.
- An anonymised online questionnaire was sent to CDM patients in the UK through CureDM and the UK DM patient registry. The questionnaire was answered by 75 patients themselves or by their carers (figure 1).
- Most of the patients documented their age, sex, nearest town, the age of onset, age of diagnosis, CTG repeats, type of inheritance, current symptoms and professionals involved with care.

Figure 1. Location of the CDM patients that answered the questionnaire.

Results

Table 1. General characteristics of the CDM patients

Age, mean (SD)		13.6 (11.97)	
		n	%
Sex	Female	31	41.3%
	Male	43	57.3%
Inheritance	Maternal	64	85.3%
	Paternal	10	13.3%
	Unknown	0	0.0%
Walking ability	Walking aids	5	6.7%
	Wheelchair use	39	52.0%
	Orthotics	42	56.0%

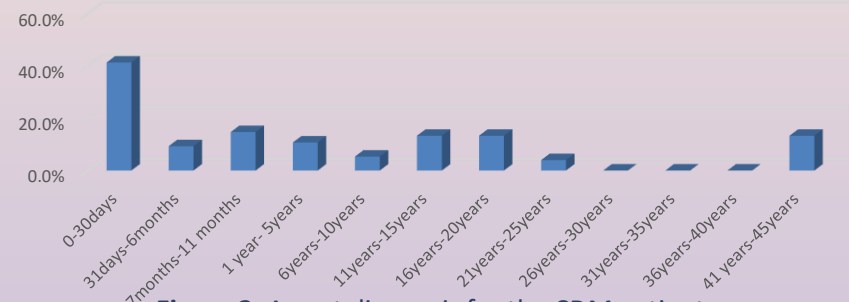

Figure 2. Age at diagnosis for the CDM patients

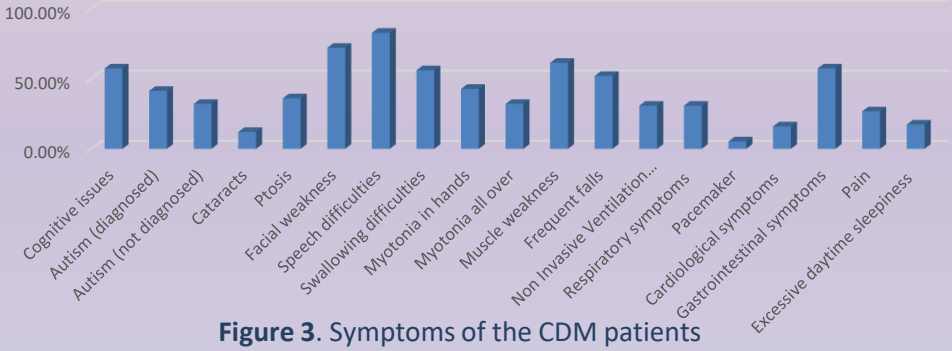

Figure 3. Symptoms of the CDM patients

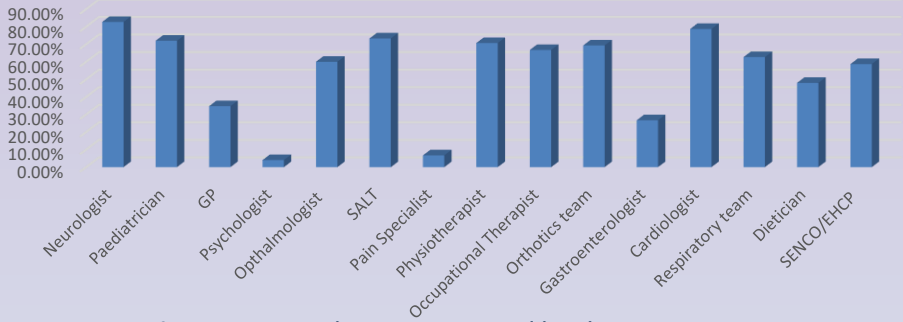

Figure 4. Specialist care received by the CDM patients

Table 2. Correlations between age and symptoms of CDM

		Uses walking stick	Uses wheelchair Splints or walking aids	SENCO / EHCP (in school)	Autism (diagnosed)	Cataracts	Ptosis	Swallowing difficulties	Myotonia in hands	Myotonia all over	Muscle weakness	Frequent falls	Respiratory symptoms	Pacemaker	Gastrointestinal symptoms	Excessive daytime sleepiness
Age	R	0.462*	0.1	0	-0.351**	0.285*	0.332**	0.415**	-0	0.466**	0.300**	0.1	0.2	-0.1	0.234*	0	.308*
	p-value	0	0.2	1	0	0	0	0	0.8	0	0	0.6	0.1	0.2	0	1	0

Conclusions

- CDM is geographically widespread around the UK, often with late or misdiagnosis. Further studies could benefit the community, leading to increased awareness, support and care, and improving quality of life.
- Gastrointestinal problems and swallowing difficulties are significant for the patients with CDM, yet gastroenterology specialist input is low. Improved awareness and support in these areas could decrease risk and improve quality of life.
- Patients report a high percentage of cognitive impairment and autism, both diagnosed and not diagnosed. However, psychologist input is underreported. Improved understanding of this symptom could support those with CDM in educational settings and increase support into adulthood.
- Paternal inheritance of CDM is often overlooked due to the maternal bias of the condition. Our study shows a paternal inheritance of 13.3% supporting the fact that it is important to educate both males and females of the inheritance risk when family planning.
- The results from this survey will help in the planning, design and recruitment for DM1 clinical trials.

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