Congenital and Childhood Myotonic Dystrophy Type 1 in the UK

Nikoletta Nikolenko[1], Ben Porter[2], Emma-Jayne Ashley[3]

1. National Hospital for Neurology and Neurosurgery, Queen Square, University College London Hospitals NHS Foundation Trust, UK

2. The John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle upon Tyne Hospitals NHS Foundation Trust, Newcastle upon Tyne, UK

3. Cure Myotonic Dystrophy UK Charity (CDM), North Lincolnshire, UK

BACKGROUND

- Myotonic Dystrophy type 1 (DM1) is the most common adult muscular dystrophy affecting 8,000 people in the UK [1]. A recent genetic study has indicated it to be far more common at 1:2100 [2]
- DM1 is a progressive multi-system autosomal dominant disorder with no disease-modifying treatment [1]. The symptoms include distal muscle weakness, myotonia, arrythmias and other multisystemic manifestations. DM1 is usually classified into 4 subtypes according to age of onset: Congenital, Childhood, Adult-onset and Late Adult [3].
- Advances in the understanding of the molecular pathogenesis of DM1 have enabled the development of a potential new targeted treatment for children, adolescents and adults with congenital and childhood onset DM1.
- However, there is limited information about the prevalence and epidemiology of paediatric onset DM1 in the UK.

AIM

To study the epidemiology of paediatric onset DM1 in the UK.

METHODS

- CureDM is a UK Charity that aims to support patients and families affected by DM1, with a particular interest in congenital and childhood onset DM1 and raising awareness for this condition.
- An anonymised online questionnaire was sent to DM1 patients in the UK through CureDM and the UK DM patient registry.
- The questionnaire was answered by the patients themselves or by their carers.

Figure 1. Location of the congenital and childhood DM1 patients that answered the questionnaire.

RESULTS (1)

75 Congenital and 107 Childhood onset DM1 patients answered an online anonymised questionnaire across the UK (**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.

Table 1. General characteristics of the Congenital and Childhood myotonic dystrophy type 1.

		Congenital onset		Childhood onset		P-value	Total
Age, mean (SD)		13.6 (11.97)		31.88 (14.38)			
Disease onset		0 years (at birth)		11.02 (5.19)			
		n	%	n	%		
Sex	Female	31	41.3%	70	65.4%	0.001	101
	Male	43	57.3%	34	31.8%		77
Inheritance	Maternal	64	85.3%	33	30.8%		97
	Paternal	10	13.3%	57	53.3%	<0.001	67
	Unknown	0	0.0%	14	13.1%		14
Walking ability	Walking aids	5	6.7%	17	15.9%	0.069	22
	Wheelchair use	39	52.0%	10	9.3%	<0.001	49
	Orthotics	42	56.0%	18	16.8%	<0.001	60

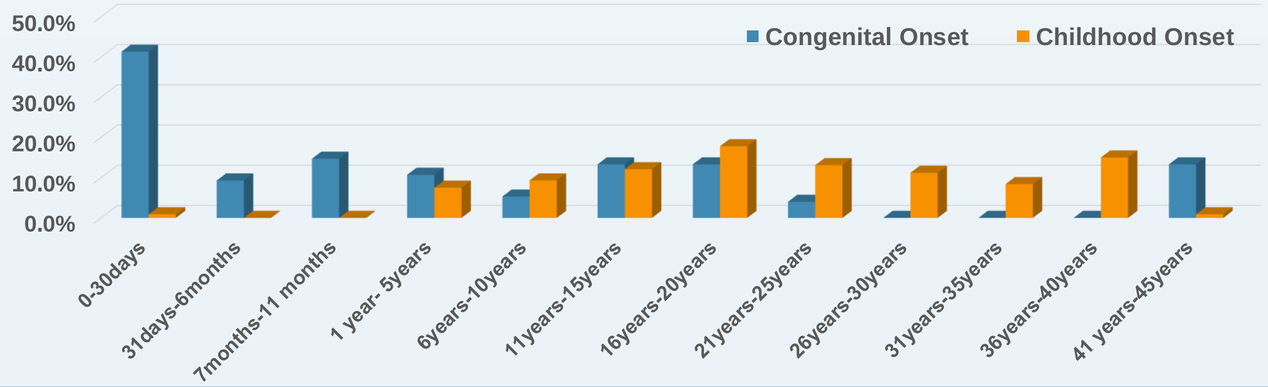

Figure 2. Age at diagnosis for the patients with congenital and childhood onset of DM1.

RESULTS (2)

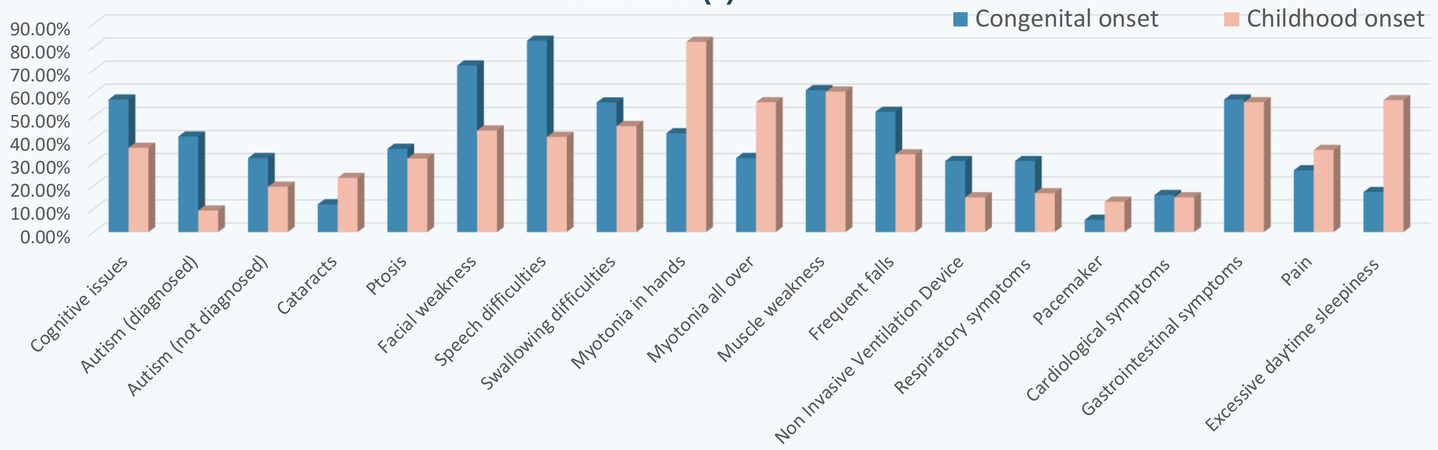

Figure 3. Symptoms for the patients with congenital and childhood onset of DM1

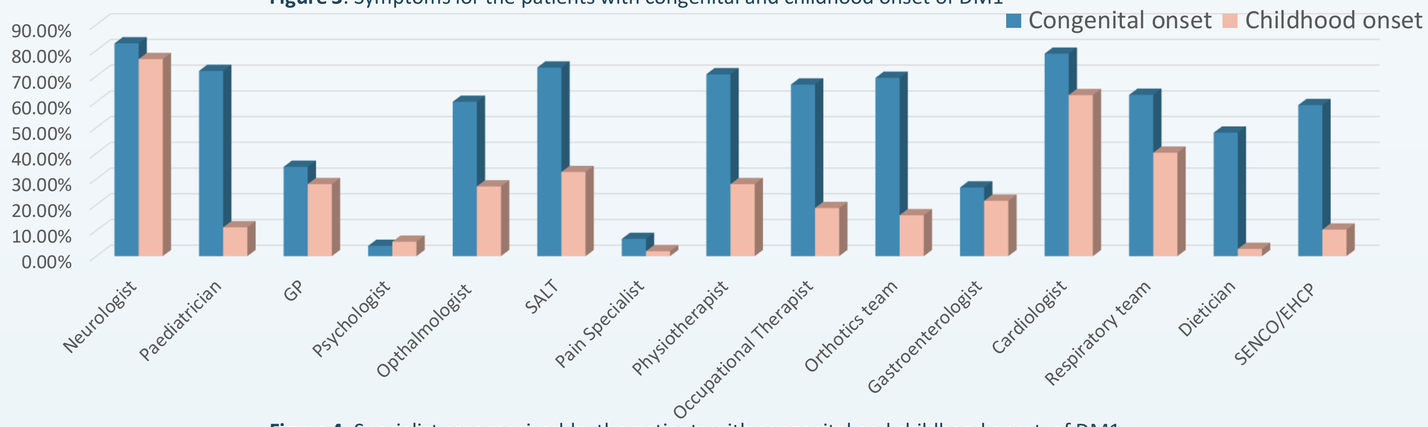

Figure 4. Specialist care received by the patients with congenital and childhood onset of DM1

Table 2. Correlations between age and age upon disease onset with symptoms of congenital and childhood onset of DM1

			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
Congenital Onset	**Age**	**R**	0.462**	0.148	0.001	-0.351**	0.285*	0.332**	0.415**	-0.036	0.466**	0.300**	0.058	0.191	-0.142	0.234*	0.004	.308**
		p-value	0.000	0.208	0.996	0.002	0.014	0.004	0.000	0.763	0.000	0.009	0.624	0.103	0.226	0.045	0.974	0.008
Childhood Onset	**Age**	**R**	0.181	0.456**	0.225*	-0.414**	-0.206*	0.458**	0.431**	0.240*	0.317**	0.290**	0.228*	0.238*	0.293**	0.495**	0.243*	0.125
		p-value	0.065	0.000	0.021	0.000	0.035	0.000	0.000	0.014	0.001	0.003	0.019	0.015	0.002	0.000	0.012	0.203
	Age upon disease Onset	**R**	0.040	.204*	0.031	-.424**	-.282**	.262**	0.108	.243*	.309**	.227*	0.092	-0.095	0.093	0.096	0.052	0.094
		p-value	0.701	0.047	0.767	0.000	0.005	0.010	0.293	0.017	0.002	0.026	0.372	0.358	0.366	0.351	0.612	0.365

CONCLUSION

Differences were noted between congenital and childhood onset DM1 in walking abilities, specific symptoms and specialist care received.

Early specialist input is key to improve the quality of life.

Genetic counselling should be provided appropriately.

The results from this survey will help in the planning, design and recruitment for DM1 clinical trials.

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CONTACT

Presenting author email address: n.nikolenko@nhs.net