New FKRP mutations causing congenital muscular dystrophy associated with mental retardation and central nervous system abnormalities. Identification of a founder mutation in Tunisian families.

Nacim Louhichi, Chahnez Triki, Susana Quijano-Roy, Pascale Richard, Samira Makri, Mériem Méziou, Brigitte Estournet, Slah Mrad, Norma B. Romero, Hammadi Ayadi, et al.

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Table 2 Louhichi et al. Neurogenetics 2004

Table 2: Summary of the clinical features of the seven patients (CK creatine kinase, ND no data)

Patient	1	2	3	4	5	6	7
Sex	F	M	F	F	F	F	M
Age of onset	Birth	Birth	Birth	4 months	2 months	Birth	Birth
Current age (years)	7	6	6	5	6	12	3
Serum CK	4,400 IU/l	5,500 IU/l	7,330 IU/l	2,978 IU/l	2,600 IU/l	4,997 IU/l	1,370 IU/l
Maximal motor milestone	Sat unsupported	Sat unsupported	Sat unsupported	Sat unsupported	Sat unsupported	Sat unsupported	Sat unsupported
Contractures	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Hypertrophy	Calf	Calf	No	Calf	No	Tongue	Calf
Mental Retardation	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Head circumference	- 2 SD	- 2 SD	- 2 SD	- 2 SD	- 2 SD	- 2 SD	- 2 SD
White Matter Changes	Yes (3 years)	Yes (3 years)	No (6 years)	No (5 years)	Yes (3 years)	Yes (1 year)/ +/-(4 years)/ No (12 years)	Yes (2.5 years)
Cerebellar Cysts	Yes	Yes	Yes	Yes	ND	Yes	Yes
Hypoplasia of vermis	Yes	Yes	Yes	No	ND	Yes	Yes
Microcephaly	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Eye involvement	No	Strabismus	No	No	No	Ophthalmoplegia	ND
Cardiac function	Normal	Normal	ND	left ventricular hypertrophy	ND	Left ventricular dilatation	Normal
Respiratory function	Normal	Normal	Normal	Normal	Normal	Mechanical ventilation	Normal
Scoliosis	Yes	Yes	No	Yes	No	Vertebral arthrodesis	No