

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

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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)**

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