

**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              |