

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)

| 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 |