investigation of these patients, incorporating both clinical and laboratory evidence. It would be of great value to carry out a skeletal muscle biopsy on the patient described by Dr ALLABAN in his histochromatographic and biochemical evidence of mitochondrial respiratory chain dysfunction. These investigations are often abnormal in patients with neurological abnormalities resulting from mtDNA defects, and the characteristic mosaic pattern of cytochrome c oxidase activity in muscle or a biochemical complex deficiency may provide clues as to the nature of the underlying genetic defect. It is often difficult to ascribe pathogenicity to a mtDNA mutation, particularly if the disease has an unusual phenotype.1 Therefore, although the clinical evidence presented by Dr ALLABAN is compatible with a mitochondrial disorder, the inference that the T4216C and G15257A nucleotide transitions are the primary aetiological factor responsible for Fuch's corneal dystrophy is unfounded.

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“Catalepsy” in Coffin-Lowry syndrome

Crow et al reported an unusual, non-epileptic, catalepsy-like phenomenon in three males with Coffin-Lowry syndrome (CLS). The authors also provided evidence of neuromuscular dysfunction as part of the phenotype by showing abnormalities on muscle ultrasound in four gene carriers, and they commented on our observation of the rapid deterioration of muscle function in two affected brothers, aged 15 and 14 years at the time of our report.1 In that report, we described the frequent occurrence of generalised epileptic seizures without pathognomonic epileptiform elements on EEG. In one of these brothers, we had occasion to follow these CBSA patients and observed that the brothers were on the threshold of the epipsychosis and had not yet had a seizure. These patients were episodic slow waves and their electro-encephalogram showed that there was a chance of having a focal seizure, as in case 1 reported by Crow et al, these episodes were precipitated by a loud noise or excitement. The frequency of these seizure-like attacks increased with age and was correlated with a further progression of the peripheral muscle wasting and of a severe thoracolumbar scoliosis, which finally resulted in acute cardiopulmonary failure.

Over the last 25 years we have had the opportunity to examine 20 other CLS males. In one of these patients, the same type of sudden, non-epileptic attacks were noted from the age of 4 years. In this patient, the attacks were precipitated by a loud noise, and there was a history of seizures in his family. In another patient, the attacks were precipitated by a loud noise and were associated with a further progression of the peripheral muscle wasting and of a severe thoracolumbar scoliosis, which finally resulted in acute cardiopulmonary failure.

In conclusion, our experience in CLS males confirms that “catalepsy” is not rare in this XLMSR syndrome, as we observed it in three of 22 male patients. The aetiology and pathogenesis of this syndrome, a collapse phenomenon remains unclear. In this perspective, it is of interest to note that these patient-like symptoms increased in frequency and severity in the two brothers, with increasing age. The occurrence of a collapse phenomenon remains unclear. It is of interest to note that these patient-like symptoms increased in frequency and severity in the two brothers, together with the occurrence of a collapse phenomenon. In conclusion, our experience in CLS males confirms that “catalepsy” is not rare in this XLMSR syndrome, as we observed it in three of 22 male patients. The aetiology and pathogenesis of this syndrome remains unclear. In this perspective, it is of interest to note that these patient-like symptoms increased in frequency and severity in the two brothers, together with the occurrence of a collapse phenomenon.