Réunion "G.E.M." (Groupe d'Etudes en Myologie) du jeudi 12 juin 2014

Auditorium, Institut de Myologie, Paris, 14h00-16h30

Résumé des observations

Equipes	Nom	Résumé	Diagnostic et/ou commentaires
Projet Myo-Capture (Jocelyn Laporte)	…	Suite à projet.	
Naveen (*Chandigarh*) Cobo, **Urtizberea** (*Hendaye*)	Purj…	Solved case. Muscle hypertrophy syndrome in an Indian child. Mildly elevated CK. No muscle biopsy available.	
Barnerias, Desguerre (*Necker*) Romero (*IDM, Paris*)	Mathilde MEND…	Hypertrophie musculaire. Biopsie musculaire non contributive.	
Testard, Meyzin (*Grenoble*)	…	SMA-like phenotype. But inconsistent pathological findings.	
Al-Rohaif, Al-Ajmi, Bastaki (*Kuwait),* Cobo, **Urtizberea** *(Hendaye)*.	…	*Case already presented in a GEM session.*Consanguineous family from Kuwait. The proband presents with a FSH-like phenotype (asymmetrical steppage, moderate scapular winging but witthout facial weakness). Mother is affected and WCB. A sister is affected. His son is asymptomatic but has weird shoulder movements.	
Olivé (*Barcelona*, Spain)	…	Muscle weakness in two adult sibs.	
Urtizberea	…	Adult-onset distal myopathy.	