Réunion "G.E.M." (Groupe d'Etudes en Myologie) du jeudi 20 juin 2013

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

Résumé des observations

Equipes	Nom	Résumé	Diagnostic retenu et/ou commentaires
Grosmann (*San Diego, USA*)	…	2 sibs. Facial and limb weakness.	FSHD ? Congenital myopathy ? Mitochondrial ? TK2 ?
Warman-Chardon (*Montréal, Canada)*	…	LGMD phenotype. 2 brothers affected. Biopsy dates back to 1983. Dystrophic pattern. Big tongue. Marked wasting. Severe dilated cardiomyopathy. All known LGMD genes are not mutated including FKRP.	New gene found via exome sequencing. Subsequent muscle biopsy is scheduled.
Laquerrière, Bedat-Millet, Guyant-Maréchale *(Rouen)*		Suspicion de myosite chez une patiente présentant un syndrome inflammatoire avec vitesse de sédimentation très augmentée. Facteur anti-nucléaire à 1/640. Bilan de myosite négatif. CPK à 1.120 unités. Sur le plan clinique, déficit des ceintures prédominant au niveau des membres inférieurs et durant depuis 12 ans. Notion de possible consanguinité. Dysferlinopathie? Mais la dysferline est normale ! Microcalcifications.	Maltase acid deficiency is one option. Check the phosphatase acid stain.
Drouin-Garraud, **Laquerrière** *(Rouen)*	…	Suspicion de myopathie avec excès d'autophagie.	XMEA or Danon disease. Check for the LAMP2 stain. If normal, screen the VMA21 gene in Finland (Udd).
Cuisset, Maurage, Renaud-Monsarrat (*Lille),* Quijano-Roy *(Garches)*, Seta, Séraphin *(Bichat)*	**Louan…M.**	Louane M., born on 3/06/2009, congenital hypotonia with limb girdle weakness, myopathic face, CPK > 15000 UI/l. Normal cerebral MRI. Muscle biopsy (17 months old) suggestive of a congenital muscular dystrophy: marked variation in fibre size, internal nuclei, extensive proliferation of connective tissue; IF: normal for merosine and collagene 6, negative for alpha-dystroglycan. Negative gene study for FKRP, Fukutin, POMT1, POMT2, POMGnT1, and LARGE. Genetic diagnosis finally resolved.	Congenital muscular dystrophy. Mutation in the ISPD gene. ISPD so far was involved mostly in Walker-Warburg syndrome.
Laforêt, Romero (*IDM)*	…	Recurrent abdominal pain, encephalopathy and high CK levels in a	MCADD. Confirmed at the molecular level.

		young adult.	
Barnérias, *Desguerre (Necker)*, Urtizberea *(Hendaye)*	Maxence B…	Suite à dossier : myopathie congénitale avec anomalies sensorielles, cas sporadique. Impasse diagnostique (cf. reportage diffusé au moment du Téléthon 2012). Nouvel élément à verser au dossier.	Significant decrement (30%). No known syndrome fits in. Exome sequencing is recommended.
Urtizberea, Cobo *(Hendaye)*, Manère *(IDM)*, Nampoothiri (*Cochin*)	Fath….	Female Indian patient aged 10 with severe proximal weakness since age 1. Elder brother deceased at 5 yr of pneumonia. Myosinopathy suspected on muscle biopsy.	Pseudomyopathic SMA. Proven at the molecular level. Coexistence of type 2 and type 3 within the same family.
Romero, Eymard (Institut de Myo, Paris)	….	Sporadic case. Unusual muscle biopsy. Radiant lesions. Quite marked on EM.	New entity ?
Kapetanovic (*Bilbao*), Gallano (*Barcelona*) **Urtizberea** (*Hendaye*)	…	Distal myopathy in a Roma Gypsy girl. Parental consanguinity. High CK (> 12.000). LL distal and proximal muscle weakness. Dystrophic muscle. Negative immunostain against dysferlin but no mutation found in the DYSF gene.	Miyoshi-type distal myopathy. ANO5 has been excluded. Consider screening DYSF more extensively (promoter region, MLPA). Blot dysferlin in monocytes and in muscle (if available).
Kapetanovic (*Bilbao)*, **Urtizberea** (*Hendaye*)	…	Adopted Romanian girl. Facial weakness, proximal weakness, big calves. Slow progression. Mildly elevated CK. Inclusions and vacuoles on the muscle biopsy. Selective muscle involvement on MRI.	Early onset myofibrillar myopathy ? check myotilin (Udd).