eases gives hope for the improvement of coordinated multidisciplinary treatment approach and difficult transition from pediatric to adult care.

"Congenital *LMNA*: special patients, special cardiac features"

Georgia Sarquella- Brugada
Hospital Sant Joan de Déu, Barcelona - Universitat de Barcelona, Spain

LMNA patients may present a wide variety of phenotypes. The congenital early phenotype, mostly related with drop-head syndrome, has special features concerning not only skeletal muscle, but especially cardiac disease. We present the early cardiac manifestations of a very young population of patients with congenital form of *LMNA*.

"Clinical and genetic characterisation of Lithuanian patients with muscle laminopathies"

Birutė Burnytė[1], Greta Senkevičiūtė[2], Aušra Morkūnienė[3], Deimantė Braždžiūnaitė[1], Algirdas Utkus[1].
1. Institute of Biomedical Sciences, Faculty of Medicine, Vilnius University Lithuania, 2. Faculty of Medicine, Vilnius University Lithuania, 3. Vilnius University Hospital Santaros Klinikos Lithuania.

Background: Emery-Dreifuss muscular dystrophy type 2 (EDMD2; OMIM #181350) is a rare muscle disease characterized by the clinical phenotype of progressive proximal muscle weakness, early-onset joint contractures, and cardiac involvement. Here we delineate clinical and genetic features in three cases of EDMD2. Methods: The study subjects were recruited retrospectively from the database of our institution. We reviewed the clinical, laboratory and molecular findings. Results: Distinct heterozygous *LMNA* missense variants were found to segregate with the clinical phenotype in three subjects (S1, S2 and S3) from three unrelated families. All *LMNA* variants had occurred *de novo* and were reported previously. The disease manifested during infancy or early childhood and the age of onset ranged from 0 to 1.2 years of age. Two subjects (S1 and S2) presented with motor developmental delay. Meanwhile, S3 demonstrated proximal lower limb weakness characterized by gait abnormalities. All patients had skeletal system deformities. Contractures were observed in S3 at the ankle site. S2 had chest wall deformity (pectus excavatum). S1 and S3 had hyperlordosis. Joint hypermobility was present in 2/3 subjects. CK levels were elevated in all subjects. S1 and S3 had respiratory system involvement characterized by obstructive sleep apnea episodes. Cardiac assessments revealed a sinus tachycardia in each subject. Conclusions: These study results showed typical clinical characteristics in children with EDMD2. Early genetic diagnosis is important for management of possible associated complications like cardiac diseases, requiring regular cardiological follow-up.

"Different disease progression velocity in two female monozygotic twins diagnosed with *LMNA*-related congenital muscular dystrophy"

Sergi Cesar[1], Estefania Martínez- Barrios[1], José Cruzalegui[1], Fredy Chipa[1], Andrés Nascimento[2], Carlos Ortez[2], Daniel Natera[2], Oscar Campuzano[3], Georgia Sarquella- Brugada[1]
1. Arrhythmia, Inherited Cardiac Diseases and Sudden Death Unit, Hospital Sant Joan de Déu Spain; 2. Neuromuscular Unit, Hospital Sant Joan de Déu Spain; 3. Genetics Center, Institut d'Investigacions Biomèdiques de Girona (IDIBGI) Spain.

LMNA-related congenital muscular dystrophy (L-CMD) is characterized by axial hypotonia, muscle weakness, joint contractures, spinal rigidity and progressive respiratory insufficiency. Life-threatening arrhythmias, initially without dilated cardiomyopathy (DCM), appear earlier than other phenotypes, leading into sudden cardiac death. We present the cardiac characterization in two female monochorionic diamniotic twins, enrolled in a comprehensive follow-up with implantable loop record with remote monitoring from 4 years of age. Twin 1 expressed earlier worsening neuromuscular impairment (weakness, gait problems) and earlier arrhythmias (multifocal atrial tachycardia -AT- at 7 years) than Twin 2. Intermittent AT started one year later in Twin 2. Both patients showed aggressive cardiac impairment, characterized by refractory multifocal AT despite pharmacological treatment. Rapidly progressive heart failure (HF) with DCM showed in both cases, leading to death at 8 and 9 years of age (respectively). AT and HF were related to right ventricular thrombus in twin 1 and a stroke in twin 2. Genetic testing showed the pathogenic variant *LMNA*: N39K of (exon 1). Additionally, other variants in other genes were identified: *CHRND*: P307S, *AGRN*: P325R and *DMD*: Q206L, all classified as having