Simposio Internacional: Enfermedades neuromusculares: es el tiempo para el tratamiento
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Diagnosis and treatment of neuromuscular junction disorders (congenital masthenic syndromes)

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Neuromuscular junction disorders, also called Myasthenic syndromes (MS), are a rare heterogeneous group of acquired (Myasthenia Gravis, MG) and inherited (Congenital Myasthenic Syndromes, CMS) neuromuscular disorders associated with distinctive clinical, electrophysiological, laboratory and ultrastructural abnormalities. The genetic defects in CMS either impair neuromuscular transmission directly or result in secondary impairments, which eventually compromise the safety margin of neuromuscular transmission. More recently, we have identified two genes (*DOK7*, *GFPT1*) that cause fatigable weakness of muscles in a limb-girdle distribution, but rarely affecting facial or eye muscles. We will cover the significant progress made in understanding the molecular pathogenesis of CMS, which is important for both patients and clinicians in terms of reaching a definite diagnosis and selecting the most appropriate treatment.