Trabalhos Científicos

Título: Delays In Time To Diagnosis Of Duchenne Muscular Dystrophy: Implications For Patient Management.

Autores: PETER RIEBLING (PTC THERAPEUTICS, NJ, USA); ABDALLAH DELAGE (PTC THERAPEUTICS, ZUG, SWITZERLAND); EDWARD O´MARA (PTC THERAPEUTICS, NJ, USA); DEMET OLESEN (PTC THERAPEUTICS, NJ, USA); PANAYIOTA TRIFILLIS (PTC THERAPEUTICS, NJ, USA); ROBERT SPIEGEL (PTC THERAPEUTICS, NJ, USA); ANDRESSA FEDERHEN (PTC THERAPEUTICS, SP, BRAZIL); TUYEN ONG (PTC THERAPEUTICS, NJ, USA)

Resumo: Introduction: Duchenne muscular dystrophy (DMD) is a severe and rare neuromuscular X-linked genetic disorder that results in functional decline, loss of ambulation and early death. Objective: To present data on current delays in DMD diagnosis. Methods: Data sources included an ongoing, post-approval, registry study (Study 025, NCT02369731) as well as two randomized controlled trials (RCT), studies 007 and 020 (NCT00592553 and NCT01826487), of patients with nonsense mutation DMD (nmDMD) receiving treatment with ataluren. An online/phone physician survey on current diagnostic and treatment practices for patients with DMD was also conducted. Paediatric or adult neurologists, or neuromuscular specialists were eligible for inclusion if they had a minimum patient threshold, had been practicing for ?2 to <35 years, and had spent ?25% of their professional time treating or managing patients with DMD. Physicians from 10 countries were included. Results: Clinical studies: In Study 025, patients' (n=78) medical histories showed that there was a mean (standard deviation [SD]) delay in time to diagnosis of 2.4 (2.3) years from age at first symptom recognition by caregiver or physician and age at diagnosis; patients were diagnosed at a mean (SD) age of 5.0 (3.3) years. The mean age at diagnosis reported in Studies 007 and 020 was 3.7 years and 4.4 years, respectively. Online physician survey: Responses showed a ~2-year delay in time to diagnosis for patients with DMD. This consisted of a ~1-year delay between time from first symptom recognition by family/caregiver to seeking advice from a healthcare professional. There was a further ~1-year delay until a confirmed diagnosis. Conclusions: There continue to be delays in the diagnosis of DMD. Initial symptoms are missed or dismissed by both families/caregivers and healthcare professionals. Reducing the time to diagnosis may allow earlier intervention, and possibly enable prolonged preservation of function.