NIHR Great Ormond Street Biomedical Research Centre

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Can treatment-related short-term neurofilament change predict long-term functional outcome in SMA?

Spinal Muscular Atrophy (SMA) is a severe, childhood-onset disease that causes muscular weakness. Children with the most severe type, SMA I are never able to sit, crawl or walk. These children historically rarely survived past the age of two years. However, three drugs have recently become available to patients thus allowing treated patients to experience prolonged survival and to achieve unexpected motor milestones. However, there is a lot of variation in how much children improve with treatment, and a lot remains unknown about why there is so much variability in response to treatment. This project aims at using novel machine learning approaches, along with biological markers called neurofilaments to understand if baseline and short-term changes in these biological measures translates to long-term functional improvement.