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ADULTHOOD TRANSITION AGE IN DYSTROPHINOPATHIES: A MANAGEMENT MODEL FOR INTEGRATED CARE DELIVERY IN TUSCANY REGION

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A multidisciplinary care approach is fundamental in neuromuscular diseases due to their complex characteristics, requiring a timely diagnosis and optimization of the management plan. Diagnostic yield has greatly improved in the last years, due either to powerful genetic-molecular techniques or increased attention of physicians to red flags that can indicate a neuromuscular disorder for prompting a specialist evaluation. The improvement of standards of care and the availability of effective disease-modifying treatments such as enzymatic replacement therapies, RNA-based and also gene therapy, but also steroid therapy for Duchenne muscular dystrophy, have expanded life expectancy of patients, also improving their quality of life, although at the same time leading to new and more complex phenotypes. It also means that, for diseases with infantile onset, greater number of patients reach adulthood, this making it necessary to establish a "transition" process from pediatric to adult care centers for structured management plans and to mitigate the risk to be lost. We have now developed in Tuscany a program for transition involving two pediatric reference centers for rare and neuromuscular disorders (IRCCS Meyer, Florence and IRCCS Stella Maris, Calambrone, Pisa) and the adult reference center for neuromuscular disorder of Pisa, University Hospital, Ospedale Santa Chiara, ERN-NMD affiliated). This program, started in 2023, includes combined and integrated evaluation of adolescent-juvenile patients from childhood neuropsychiatrists and pediatricians and adult neurologists who, in exchanging their expertise, accompany affected people in this comprehensive journey for ensuring suitable standards of care. In total, 70 neuromuscular patients transitioned from the pediatric centers to the adult center, among them 11 affected by Duchenne muscular dystrophy and 30 by Becker muscular dystrophy, for which clinical and investigation data have been extensively collected for better understanding of disease natural history and responses to interventions and treatment.

Keywords: *Dystrophynopathies, Duchenne and Becker Muscular atrophy, adulthood transition age.*