Transition from pediatric to adult care: one-year experience from a national reference centers for Neuromuscular diseases network

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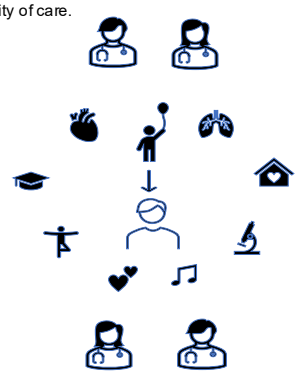
BACKGROUND

Neuromuscular disorders represent a complex and heterogeneous family of rare diseases, requiring a timely diagnosis and optimization of the management plan, which is multidisciplinary in most cases. Diagnostic yield has greatly improved in the last years, both due to the increasingly powerful and sensitive genetic analysis techniques and a greater attention of physicians to signs and symptoms that may suggest a neuromuscular disorder and prompt a specialist evaluation. Moreover, the presence of newborn screening tests for some of these diseases, as Pompe disease and Spinal Muscular Atrophy (SMA) has contributed to a number of precocious diagnoses. In Italy, these screenings are available in most of the country and are planned to be extended nationally. Moreover, the improvement of standards of care and the availability of effective, disease-modifying treatments - as enzymatic replacement therapies, SMA drugs and steroidal therapy for Duchenne muscular dystrophy (DMD) - has expanded patients' lifespan and improved their quality of life, although also contributing to the development of new and more complex phenotypes. This translates into a higher number of subjects reaching the "transition" phase to the adult care centers from the pediatric ones and suggests the need for structured management plans that guarantee a smooth passage and lowers the number of patients that may be lost at follow-up. With this concept in mind, the Italian region of Tuscany developed a program for transition from two reference centers for pediatric rare and neuromuscular disorders (IRCCS Meyer and IRCCS Stella Maris) to the adult reference center (University Hospital of Pisa).

METHODS

Patients were assessed in two outpatients visits, the first one in the pediatric center in presence of the adult neurologist and the second one, 6 months later regardless of the diagnosis or earlier if needed, in the adult center. Retrospective data on the clinical and genetic diagnosis, muscle biopsy, cardiac and respiratory function and disease-specific comorbidities were collected. A multidisciplinary team of adult specialists (cardiologists, pulmonologists etc.) was also developed to ensure global continuity of care.



RESULTS

In total, in the considered period, 70 patients (9 females, 61 males) transitioned from the pediatric centers (IRCCS Meyer and IRCCS Stella Maris) to the adult center (University Hospital of Pisa), aged 18-45 years. Most of the patients had previously received a genetic and clinical diagnosis, while 6 were asymptomatic or paucisymptomatic hyperCKemias without a definite genetic diagnosis (Fig. 1). All patients were firstly seen in the pediatric center in presence of the adult neurologist and pediatrician/childhood neurologist and then re-evaluated after 6 months at the adult center; after that visit, the ordinary schedule of follow-up was re-established according to the disease progression and patients' needs. Regarding multidisciplinary care, 52 patients were referred to specialized adult services for cardiology (BMD, DMD and other myopathies), 12 to pulmonology (DMD, SMA, LOPD) and 6 to rheumatological evaluation for bone health management (DMD) (Fig. 2). Other specific needs (i.e. nutritional care, speech therapy and swallowing assessment etc.) were addressed based on the disease. Only one patient (LOPD, already 29 y.o. at the time of transition) was regularly assessed by a psychologist, which remained unchanged.

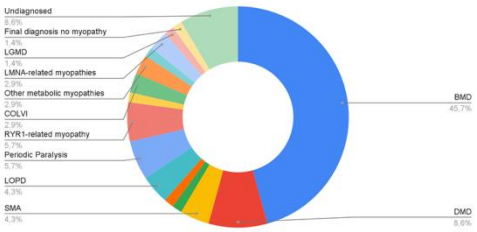


Fig.1 Composition of the study population

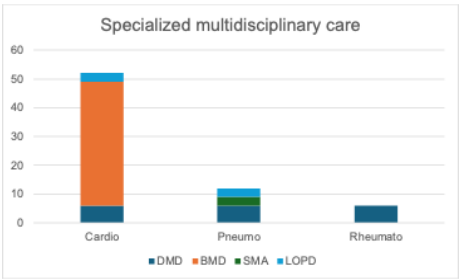


Fig. 2 Multidisciplinary care referrals in different diseases

CONCLUSIONS

The transition phase from pediatric to adult care for rare disease must encompass a coordinated action involving the pediatric and adult neurologists and all the multidisciplinary care specialists involved. Our experience outlines the compleity and numerosity of the population of interest and the need for a planned intervention.