Developmental delay in Duchenne Muscular Dystrophy: A case-control study of a Dutch population

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Background and aims: Diagnostic delay often occurs in the treatment of Duchenne Muscular Dystrophy (DMD). As the early course of the disease is defined by developmental delay, detecting this delay may aid timely diagnosis. Our aim is to investigate the differences in attainment of developmental milestones, medical referrals and symptoms between boys with DMD and boys from the general population.
Methods: Within the 4D-DMD study (Detection by Developmental Delay in Dutch boys with Duchenne Muscular Dystrophy), data on the attainment of milestones, medical referrals, and symptoms in boys with and without DMD were extracted from the (electronic) health records of the Youth Health Care Service. Additional information for boys with DMD was acquired from questionnaires.
Results: In total, health records from 76 boys with DMD and from 19,086 boys from the general population without DMD, and questionnaires from 71 parents of boys with DMD were retrieved. The presence of developmental delay became evident shortly after birth, at 2-3 months of age with a lower proportion of DMD boys attaining milestones of fine and gross motor activity (Odds Ratios (OR) ranged between 3.3 and 3.6, $p<0.001$). Between 12 and 24 months of age, differences in attainment of milestones concerning the gross motor became greater between the DMD group versus the control group activity (OR ranges: 10.6-82.4, $p<0.001$). Also, smaller, but significant differences were found in the milestones concerning fine motor activity, adaptive and personal/social behavior and communication between 12 and 48 months of age (OR ranges: 3.1-6.0, $p<0.001$). Before the diagnosis of DMD, boys with DMD were referred more often to physical therapy, speech-language therapy, an ENT-specialist and preschool educational intervention compared to the control group. Symptoms that appeared more often in DMD boys compared to peers were falling more frequently, stiff way of walking, a younger appearance than his chronological age and pseudohypertrophy.
Conclusions: Our study found substantial differences in milestone attainment, medical referrals and symptoms in boys with DMD compared to controls, which were observable from a young age.
Main question: How can we use this information within youth health care for the early detection of boys with DMD?

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Desirability of neonatal screening for Duchenne Muscular Dystrophy and Pompe Disease in Flanders.

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Background: The neonatal Guthrie test has been used in Belgium since 1968 for the detection of 11 treatable metabolic and hormonal diseases. Cystic fibrosis was added in 2019. Two other conditions considered for screening are Duchenne and Pompe disease, which both result in progressive muscle deterioration. Although technically possible, screening for these diseases has advantages and disadvantages and not all criteria for screening (as postulated by Wilson and Jungner) are met. We investigated parental acceptability of such screening extension as a feasibility criterium, and explored the position of key neurologists involved in treatment.
Methods: Parents of healthy infants (<12 months age) in the maternity ward of three hospitals and in waiting rooms of 'Child and Family' well-baby clinics in Flanders were invited to complete a questionnaire about screening for either Duchenne or Pompe disease. The results were analysed using inferential statistics (SPSS Statistics 24). Similar questionnaires were sent to (parents of) patients with Duchenne or Pompe disease in Belgium and analysed using descriptive statistics. Semi-structured interviews were conducted with neurologists of three Flemish Neuromuscular Reference Centres.
Results: In total 377 surveys were completed by Flemish parents of healthy infants. 125/148 (84.5% (95%CI 77.4-89.7%)) supported screening for Duchenne disease. 197/224 (88.0% (95%CI 82.8-91.8%)) were in favour of screening for Pompe disease. Early treatment was considered the main benefit of screening.
In total 63 (parents of) patients completed a questionnaire of whom 34/38 (89.5%) of (parents of) patients with Duchenne disease supported screening for their disease, and 24/25 (96.0%) of (parents of) patients with Pompe disease. Their main reasons to opt for screening were early diagnosis (for Duchenne) and early treatment (for Pompe). The neurologist's opinions were diverse.
Conclusion: This survey indicates neonatal screening for Duchenne and Pompe disease is largely supported by parents of newborns and (parents of) patients in Flanders. The possibility of early diagnosis and treatment were the main arguments to opt for screening.
Statement: Pompe and Duchenne disease are considered for neonatal screening. Early diagnosis allows genetic counselling, prenatal diagnosis and early treatment. The population being in favour of screening is a valuable argument in the decision process.

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Socioeconomic and ethnic differences in lung function and asthma at school age: the Generation R Study

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