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Early neurodevelopmental findings in young children with Duchenne muscular dystrophy
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Cognitive abilities have been extensively studied in children and adults with Duchenne muscular dystrophy (DMD) but less has been reported on younger children. This is not surprising as the diagnosis of DMD remains at a mean age of 4 and 6 months. The aim of this study was to assess neurodevelopmental abilities before the age of 4 years in a cohort of DMD patients in whom early diagnosis was possible because of incidental raised creatine kinase levels, family history or neonatal screenings. Sixty-six children with a diagnosis of DMD of age between 7 and 43 months (mean 27 months) were included in the study. All children were assessed using the Griffiths Mental Development Scales, establishing both total scores and subscores for each of the subscales. Thirty-six of the 66 children had a developmental quotient (DQ) > 85, 21 had a DQ between 70 and 84 and 9 below 70. The mean total DQ was 86 (SD 16.07) and the mean scores for each subscale were as follows: (A) locomotor: 79 (19.47), (B) personal social: 91 (18.48), (C) hearing and speech: 85 (23.59), (D) eye hand coordination: 87 (17.20), (E) performance: 88 (18.40), (F) practical reasoning: 95 (16.08). (Scale F only for patients older than 2 years.) Details of mutations were available in 58 of the 66 children included. Low DQ were found in five of the 26 (18.5 %) patients with mutations before exon 44, in 15 of the 29 with mutations between 44 and 55 (51.7 %) and in all three with mutations in the exons beyond exon 55. Conclusions: The DQ found in our cohort was on average one SD below a DQ of 100 with approximately 45% of the children having a DQ below 85. The locomotor scale had the lowest scores but even when we excluded the locomotor scale, a DQ below 85 in 28 patients (43%).

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S.P.57
Duchenne muscular dystrophy and epilepsy
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The aim of this study was to establish a possible present or past history of epilepsy in patients affected by Duchenne muscular dystrophy (DMD). We also performed EEG in all the boys in whom there were episodes described as ‘staring episodes’, tremors, or other abnormal movements suggestive of seizures. We report the data obtained in 217 DMD patients with proven mutations from three tertiary neuromuscular centers in Italy. Age at onset of seizures, type of seizures, family history, treatment, EEG, brain MRI (if available) were recorded. Associated cognitive and neuropsychiatric disorders were also noted. Epileptic seizures were found in 14 of the 217 DMD patients (6.4%). The age of onset ranged from 3 months to 17 years and in approximately half of the cases the first seizure was reported after the age of 10. In two cases seizure were not reported by the families but, when interviewed, they reported episodes of ‘lack of contact’. The prevalence of epilepsy in DMD was therefore higher than in the general pediatric population, where is reported to be between 0.5% and 1%. The presence of cognitive impairment did not appear to increase the risk of epilepsy and although seizures were more frequent in the subgroup of patients with mutations between exons 44 and 54, there was not a clear correlation with site of mutations as observed for cognitive impairment.

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S.P.58
Assessment of oromotor functions of children with Duchenne muscular dystrophy
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Weakness and wasting of skeletal muscles in DMD primarily cause immobility and respiratory insufficiency. Orofacial, oropharyngeal and gastrointestinal tract muscle groups are involved in the deteriorative process. Aim of the study was to analyze the correlation of influenced oromotor and other body components between feeding and swallowing disorders in DMD patients. Twenty-five subjects (mean age 10.52 ± 3.37 years) have been included to our study. To compare the chewing performance; a control group which consist of 25 healthy subjects (mean age 11.04 ± 1.72 years) was also included to study. All the participants were 5 years and/or older age, fed normally (oral) and independently. First of all, the detailed anamnesis was taken from the patients and “Feeding Profile Questionnaire” was filled by parents. Afterwards, functional levels were evaluated by Brooke & Vignos scales. Following the procedure; clinical rating scale for head control, IOP1 for tongue strength, SAFE for swallowing function, OSMSE-3 for peripheral oral speech mechanisms were assessed. To interpretate the chewing performance, “Chewing Video Analysis” was done to both of the groups. Fourteen patients (56%) were indicating physical examination findings regarding swallowing disorders and 13 patients (52%) were indicating for oral phase disorders. It was found that; general functional state effects swallowing function (it p < 0.05) and poor head control effects oromotor functions (it p < 0.05). Tongue elevation strength and swallowing ability; deteriorates with increasing age (it p < 0.05). The chewing period of subjects with DMD was found extended in comparison with healthy subjects (it p < 0.05). Oromotor disorders can be seen even though the patients are at early stage since the muscle weakness appears in DMD. These oromotor and swallowing disorders can be based onto head-trunk incapacity.

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S.P.59
Current care practice in Duchenne Muscular Dystrophy in Europe – results of the CARE-NMD cross-sectional survey
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The aim of this study was to establish a possible present or past history of epilepsy in patients affected by Duchenne muscular dystrophy (DMD). We also performed EEG in all the boys in whom there were episodes described as ‘staring episodes’, tremors, or other abnormal movements
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Neurobehavioural disorders in Duchenne Muscular Dystrophy

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Variable neurobehavioural disorders and IQ one SD below average are recognised comorbidities in Duchenne muscular dystrophy (DMD), reflecting the disrupted expression of different length dystrophin isoforms in the brain, based on the dystrophin gene mutation site. Following a screening of 84 DMD boys with validated questionnaires, we previously reported 15 with severe learning disability and 41 with scores predictive of autistic spectrum disorder (ASD). Attention deficit and hyperactivity disorder (ADHD) traits, conduct and emotional problems were also described. Overall, a higher proportion of boys affected had mutations towards the 3’ end of dystrophin. We performed targeted neuropsychological assessments including: Wechsler Intelligence Children Scale-IV (WISC-IV), Developmental Dimensional and Diagnostic Interview (3Di), Conners-3 Questionnaires, Child Behaviour Checklist (CBCL).

We found marked unevenness of performance on the WISC-IV. Eight out of 12 boys had significant difference between verbal comprehension (VCI) and perceptual reasoning (PRI), with VCI more compromised. Eight out of 19 met criteria for ASD on the 3Di. In 5 boys with ASD the CBCL showed higher scores for internalising and externalising difficulties compared to non-ASD. There were associations between lower IQ and autistic social communication difficulties. On the Conners 6/17 boys met criteria for hyperactivity and 7 for inattention problems. There was a strong association between ASD and severe ADHD symptoms of hyperactivity (OR = 22.5) and inattention (OR = 54). There was a trend towards children with mutations towards the 3’ end of the gene, having a greater chance of ASD (OR = 3.4). In our on-going study, neurobehavioural disorders emerged as important facets in DMD, with confirmed ASD prevalence rates much higher than the general population. Whilst further exploring the role of dystrophin in the brain, children with DMD should be provided with neurobehavioural-targeted support.

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S.P.61

Recurrent pseudo-obstruction and sigmoid volvulus in Duchenne Muscular Dystrophy: A case report

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Dystrophin is expressed in the smooth muscle of the gastrointestinal tract (GIT). In Duchenne Muscular Dystrophy (DMD) a disruption of protein expression can lead to functional disturbances of the GIT including acute gastric dilatation, gastroparesis and intestinal pseudo-obstruction. We report a case of a 24 year old young man with DMD confirmed by a nonsense mutation of intron 44, who developed sigmoid volvulus requiring multiple surgical interventions. Our patient presented with a long-standing history of abdominal bloating and constipation. These episodes, which were initially attributed to the use of night-time BiPAP, progressed into acute abdominal distension with visible large loop of bowel and peristalsis. He had no preceding history of vomiting or abdominal pain. Abdominal radiography showed a dilated sigmoid colon with features suggestive of volvulus. He had recurrent episodes of pseudo-obstruction over the course of two months, requiring several emergency sigmoidoscopy procedures to decompress obstructed bowel. An abdominal CT scan with contrast confirmed significant pancolonic dilatation with features of sigmoid volvulus. The post-operative course was complicated by an ileus necessitating a third emergency laparotomy with drainage of the peritoneum. Post-operatively he remained in ICU fully ventilated with a tracheostomy in situ and total parenteral nutrition. Echocardiography showed a moderately impaired systolic function. Our case illustrates that GIT functional disturbances may be serious sequelae in DMD. Non-invasive ventilation and use of steroids can be confounders for underlying GIT dysmotility. Young adults with DMD, who present with