

ABSTRACT

Background : Duchenne Muscular Dystrophy (DMD) is a rare genetic neuromuscular disorder caused by mutations in the dystrophin gene. The progression of the disease is expected to negatively influence the patients' health-related quality of life. Aside from physical disability, low quality of life was expected to be influenced by cognitive disabilities found in DMD patients caused by the expression of dystrophin genes in the brain. This statement is supported by previous studies that reported the negative influence of intellectual disability on the quality of life in patients. However, the relationship between the cognitive profile and health-related quality of life in DMD patients was never previously studied. There is also no previous record in Indonesia on quality of life in DMD patients.

Objective: The purpose of this study was to assess cognitive profile, quality of life, and the correlative relationship between the two variables in DMD patients in Yogyakarta

Method : A total of 23 DMD patients aged 5 – 18 registered in RSUP during 2017 – 2020 were involved in the study. Cognitive function was calculated using Wechsler Intelligence Scale for Children – Fifth Edition (WISC-V) and quality of life was measured using the Pediatric Quality of Life 4.0 (PedsQL) Generic Module, which divides total, physical, emotional, social, and school functions. Quality of life was compared between the child's perspective (HRQoL child self-report) and parents' perspective (HRQoL parent proxy-report). Correlation between cognitive profile and quality of life in DMD patients was analyzed using the Pearson correlation test. Value of p less than 0,05 is considered significant statistically.

Result : A significant relationship was found between cognitive profile and social function of HRQoL child self-report ($\rho = 0,468$; p-value = 0,028). The cognitive profile of DMD patients was presented in mean IQ of 77,86 ($\pm 20,42$), which was lower than the average IQ score of children in Yogyakarta. Mean total score of HRQoL child self-report was 66,16 ($\pm 23,36$) with lower score in physical (IQR = 66,16) and social function (mean = 68,7). Mean total score of HRQoL parent proxy-report was higher with the score of 72,34 ($\pm 25,09$) and also lower scores in physical (IQR = 78) and social function (mean = 70). Significant relationships were also found in HRQoL child self-report and some confounding variables, including patients' age, mother's education, and walking ability. HRQoL parent proxy-report was also significantly correlated to patients' age and walking ability.

Conclusion : Cognitive profile in DMD patients is significantly correlated to their health-related quality of life, although the relationship is only found in social function from the child's perspective. The cognitive profile shown in the mean IQ of 77,86 is lower than the average. The mean total score of HRQoL child self-report of DMD patients was 66,16, while HRQoL parent proxy-report has a mean total score of 72,34. HRQoL also has a significant relationship with age, mother's education, and patient's walking ability.

Keyword(s) : Duchenne Muscular Dystrophy (DMD); health-related quality of life; cognitive profile

ABSTRAK

Latar Belakang : *Duchenne Muscular Dystrophy* (DMD) adalah penyakit genetik neuromuskular langka yang disebabkan oleh mutasi gen distrofin. Progresi penyakit dapat menjadi salah satu faktor negatif yang mempengaruhi kualitas hidup pasien. Selain disabilitas fisik, rendahnya kualitas hidup pasien juga dapat dipengaruhi oleh gangguan kognitif pada pasien DMD yang disebabkan karena terganggunya ekspresi gen distrofin di otak. Pernyataan ini didukung oleh beberapa penelitian sebelumnya yang melaporkan hubungan negatif antara disabilitas intelektual dengan kualitas hidup pasien. Namun, hubungan antara profil kognitif dan kualitas hidup terkait kesehatan pasien DMD belum pernah diteliti sebelumnya. Sejauh ini juga belum ada terkait kualitas hidup pasien DMD di Indonesia.

Tujuan: Mengetahui data profil kognitif serta kualitas hidup pasien DMD serta hubungan profil kognitif dengan kualitas hidup pasien DMD di Yogyakarta.

Metode : Subjek penelitian adalah pasien DMD berusia 5 – 18 tahun di RSUP Dr. Sardjito tahun 2017 - 2020. Fungsi kognitif pasien diukur menggunakan *Wechsler Intelligence Scale for Children – Fifth Edition* (WISC-V). Kualitas hidup pasien DMD diperiksa menggunakan *Pediatric Quality of Life 4.0 (PedsQL) Generic Module* dan disajikan dalam nilai total beserta fungsi fisik, sosial, emosi, dan sekolah. Hasil kualitas hidup juga dibedakan berdasarkan penilaian anak (*HRQoL child self-report*) dan penilaian orangtua (*HRQoL parent proxy-report*). Korelasi antara profil kognitif dan kualitas hidup pasien DMD diperiksa menggunakan tes korelasi Pearson. Nilai p kurang dari 0,05 dianggap signifikan secara statistik.

Hasil : Hubungan signifikan ditemukan pada IQ dan fungsi sosial *HRQoL child self-report* ($p = 0,468$; $p\text{-value} = 0,028$). Profil kognitif pasien DMD ditunjukkan dalam mean IQ sebesar 77,86 ($\pm 20,42$) yang lebih rendah dibandingkan rerata IQ anak di Yogyakarta. Rerata nilai total *HRQoL child self-report* adalah 77,86 ($\pm 23,36$) dengan nilai lebih rendah pada fungsi fisik (IQR = 66,16) dan sosial (mean = 68,7). Sementara itu, *HRQoL parent proxy-report* memiliki rerata lebih tinggi sebanyak 72,34 ($\pm 25,09$) dengan nilai yang lebih rendah pada fungsi fisik (IQR = 78) dan sosial (mean = 70). Hubungan signifikan juga ditemukan pada *HRQoL child self-report* dengan beberapa variable perancu, yaitu usia pasien, pendidikan ibu, dan kemampuan berjalan. *HRQoL parent proxy-report* juga memiliki hubungan signifikan dengan usia pasien dan kemampuan berjalan.

Kesimpulan : Profil kognitif memiliki hubungan signifikan dengan kualitas hidup, meskipun hubungan ini hanya ditemukan pada fungsi sosial dari *HRQoL child self-report*. Profil kognitif pasien DMD ditunjukkan pada rerata nilai IQ sebesar 77,86. *HRQoL child self-report* pasien DMD memiliki rerata nilai total 66,16. *HRQoL parent proxy-report* memiliki rerata nilai total 72,34. *HRQoL* juga memiliki hubungan signifikan dengan usia, pendidikan ibu, dan kemampuan pasien berjalan.

Kata kunci : *Duchenne Muscular Dystrophy* (DMD); kualitas hidup terkait kesehatan; profil kognitif