Case Report

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Case Report

Late Consequences of Lifting Heavy Weights by an Individual with Belated Diagnosis of Marfan Syndrome

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Abstract: Introduction: Marfan syndrome (MS) is a genetic disorder with weakening of connective tissue, tall stature, thinness, arachnodactyly, long arms and legs. Clinical manifestations may include a wide range of cardiovascular, ocular, and musculoskeletal derangements. The case presented here is unique because it describes consequences of belated diagnosis of moderately expressed MS. Such cases are probably often overlooked and underreported. Case Presentation: This case reports a 68-year-old patient with symptoms of MS and corresponding family history, who presented with progressing prolaps of lower abdominal wall, perineal descent, femoral hernia and foot edema on the site of the hernia. MS was diagnosed at the age of over 30 years. Before that, the patient worked in construction and was engaged in other activities, including lifting of heavy weights. Apart from wearing a bandage, no therapeutic interventions have been implemented so far. Conclusion: Patients with MS should avoid heavy weight lifting. Importance of timely diagnosis of MS and genetic counseling of individuals at risk is stressed.

Keywords: marfan syndrome; heavy weightlifting; abdominal wall prolapse; hernia; genetic counseling; inguinal region

1. Introduction

Marfan syndrome (MS) is a disorder with weakness of connective tissue, inherited according to the autosomal dominant pattern, having variable penetrance [1,2]. There is abundant literature about MS; and the symptoms are generally known. This case report illustrates late consequences of lifting heavy weights by an individual with belated diagnosis of moderately expressed MS. Patients with MS are discouraged from heavy weightlifting, contact sports (rugby, boxing), high G-force activities and deep-sea diving [3]. However, most of them may engage in athletic and other physical activities with moderate load. At the same time, it was noticed that there is little real-world data to support the lifestyle recommendations [1].

2. Case Report

The family history of S. illustrated the psychological mechanism "like will to like", which can facilitate marriages between people with related hereditary conditions. Other people would perceive an abnormality, but a person somewhat similar to oneself might provoke interest and sympathy. This is potentially dangerous for the offspring. Both his parents were ethnic Russians. The mother had moderately expressed marfanoid appearance. The father was tall with the head circumference ~61 cm, otherwise nondescript. However, the father's sister and daughter from his first marriage both had marfanoid body structure. S. had moderately expressed symptoms of MS: tall stature, thin skeleton with lax joints, moderate arachnodactyly (Figures 1 and 2), pes planus and slight kyphosis diagnosed in childhood. From adolescence on, S. heard comments about his "feebleness" and overcompensated this by some sports: rowing, backpacker tourism, fitness training including weightlifting. Starting at the age of 18 years, he repeatedly participated in construction and forestry

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works during holydays, totaled to 3 years plus 2 years of army service. These activities included lifting of heavy weights. Marfan syndrome was not diagnosed at yearly checkups (dispensarizations) neither at the Sechenov Medical Academy, where S. studied and was employed in the period 1973-1990, nor at other institutions. Hereditary diseases have not been given sufficient attention in the above-named Academy [4]. The diagnosis was first suspected at the age over 30 years, when S. already had visible asymmetry of the inguinal region. At the age of 66 years, along with atrophy of abdominal muscles, S. developed prolapse of his lower abdominal wall and femoral hernia with foot edema on the same left side (Figures 1 and 2). He can sleep only keeping his lower limbs apart e.g. with a balloon ~40 cm in diameter between his thighs, otherwise circulation disturbances in the limbs become perceptible due to perineal descent with compression of blood vessels. Besides, he has a hiatal hernia and radiologically confirmed old compressive fracture of the C6 vertebral body with recurrent pains and symptoms of vertebrobasilar insufficiency. Emergence of the fracture coincided with heavy weight lifting during construction works at the age of 29 years.

Figure 1. Patient S. 68 years old. Prolaps of abdominal wall, descensus perinei, femoral hernia.

Figure 2. The same patient. Increasing foot edema on the site of the femoral hernia.

3. Discussion

The vertebral injury mentioned above should be further commented. In 1985-1986 the construction job was helping to finish a stadium in a city north of St. Petersburg. Temporary workers were employed because of the short construction period, for which the regular staff did not suffice. They worked on average 16 hours a day without holidays, making concrete works and doing other tasks. The vertebral column sent feedback through the afferent channels: he should not carry heavy weights. The barrow became heavier, and the pain in the spine got worse. It started to hurt again 12 years later, in the cervical area, when S. practiced abroad as a pathologist and started to go on long-distance bicycle trips to counteract the sedentary lifestyle. At first, it was difficult to turn his head changing traffic lanes; the pain later became almost permanent, forcing him to get off the bicycle and walk. The conclusion, after radiography, was: old compressive fracture of the C6 vertebral body. S. had to avoid any load on the shoulder girdle… The barrow was overloaded by the team-leader (Dmitrii Iosifovich Gotlib). S. had a visibly thin skeleton; the barrow was too heavy for him, but he had difficulties with saying “No”; and surrounding persons knew this; more details are in the article [4]. Gotlib had pectus excavatum and treated epilepsy, which he concealed from his wife (divorced after a nocturnal grand mal) and the Khrzhizhanovsky Power Engineering Institute, where he worked with sources of radiation (dismissed in the 1990s, later worked in construction).

Many cases are known when heritable disease was concealed from the partner and then acquired by the offspring. For example, 20-25% people from certain East- and mid-European Jewish descent carry certain disease-causing genes. Approximately one in 10-15 Ashkenazi Jewish individuals are carriers of a mutation causing type 1 Gaucher disease, 1/30 – familial dysautonomia, 1/75 – A or B type of Niemann-Pick disease; 1/40 are estimated to be carriers for Canavan disease, 1/89 - Fanconi anemia and so forth [5]. These and other relevant conditions are presented in the comprehensive handbook [5]; more details with references are in the article [6]. Screening of the European Jewish population for recessive disease-causing mutations is recommended [7–10]; one grandparent suffices to offer a genetic examination [7]. Israel plays an important role in the development of genetic counseling: more than 10,000 people are tested there every year. The Zionist eugenics was designated as a prenatal policy backed by genetic technologies [11]. A motivation for eugenics has been prevention of hereditary degeneration in the human stock [12]. “Do not have children unless you are sure that they will be healthy both mentally and physically” [11]. This suggestion is not realizable in conditions of sexual and reproductive coercion, contraceptive sabotage or concealment of a heritable disease from the partner or spouse.

Many mutations are not specific but tend to be accumulated by the above-named population [13,14]. The problematic heredity may give rise to conscious or subconscious motives to look for

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partners from different ethnic backgrounds. Various tools can be applied: seduction and persuasion, intimidation and force, alcohol and drugs. Women should be aware of these tactics. The sexual and reproductive coercion has been used for the purpose of migration, to cement relationships and marriages, to obtain a residence permit and lodging, or to spread a certain genotype often with geopolitical motives. This is a probable cause of increased birthrates immediately after immigration [15].

Another case observed by the author: a young individual of Jewish descent (Aleksandr Kantsedikas, lithuanization of the name Kohen-Tsadok), who wanted to live in Moscow, concealed a hereditary condition from his older ethnic Russian bride. He had a male Stein-Leventhal alias polycystic ovary syndrome (PCOS) equivalent, possibly combined with other derangements: gynecomastia and hypertrichosis, polythelia and inclination to overweight. Male PCOS was defined as a syndrome with signs of hyperandrogenism, PCOS-like metabolic pattern and familiar history of PCOS [16,17]. Their daughter inherited PCOS and died in her fifties. A vice versa case was also observed: a female postgraduate student of Baltic origin with marfanoid body structure married a Jewish department head; both their children inherited marfanoid appearance. Another example: a young male concealed type 1 diabetes mellitus from his older ethnic Russian bride; their daughter developed the same condition plus obesity at the age of ~25 years, when her father had already been dead.

4. Conclusion

MS has variable penetrance [1,2]. This means that there are many people with moderately expressed symptoms; but their connective tissue is weakened [18]. Furthermore, individuals with marfanoid phenotype may be carriers of disease-causing genes, needing genetic counseling. This is of particular importance when a person with marfanoid features finds a life partner with a similar stature. Not all of such individuals should be diagnosed with MS, but they need an examination and advice concerning occupational choice, physical activities and procreation. Most importantly, patients with MS must avoid heavy weight lifting. Genetic counseling should be broader applied in high-risk groups. Screening of Ashkenazi population for disease-causing mutations is recommended; one grandparent suffices to offer a genetic examination [7]. More attention should be given to hereditary conditions at medical checkups and in the course of medical education [19].

Conflict of Interest: The author declares no conflict of interest.

Consent for Publication: The consent has been obtained.

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