chromosome with a mutation in the FVIII gene. In addition, it is estimated that there are 1.6 female carriers for every male HA patient. HA Carriers are classified into five categories based on residual plasma FVIII level (FVIII:C) and phenotype: (i) FVIII:C > 40% with bleeding phenotypes are classified as symptomatic; (ii) FVIII:C > 40% without bleeding phenotypes are classified as asymptomatic; (iii) FVIII:C > 5% to 40% are classified as mild HA; (iv) FVIII:C 1% to 5% are classified as moderate HA; and (v) FVIII:C < 1% are classified as severe HA. The aim of this study was to analyze the clinical and hemorrhagic profiles of female relatives of patients with severe HA with intron-22 inversion treated at Fundação Hemominas and older than 12 years. To date, sixteen participants have been enrolled in the study after signing an informed consent form. Participants answered two questionnaires: 1) evaluation of bleeding - ISTH (BAT-ISTH); and 2) evaluation of arthralgia. Peripheral blood samples were collected in a citrate tube and processed for DNA extraction. Genotyping to identify intron 22 inversion was performed by inverse PCR. Of the 16 participants, seven (44%) were identified as carriers of the mutation. Among the carriers, the median (MED) age was 35 years (IQR 34 - 35.5) and the bleeding score MED was 6 (IQR 4.8 - 9). In addition, carriers reported pain in the joints of the shoulders (3 – 42.9%), hips (2 – 28.6%), knees (2 – 28.6%), and ankles (2 – 28.6%). For participants who were not carriers, the age MED was 36 years (IQR 30 - 56) and the bleeding score MED was 4 (IQR 1 - 7). In addition, participants who were not carriers had pain in all joints studied, i.e., shoulders (3 – 33.33%), elbows (1 – 11.11%), hips (2 – 22.22%), knees (4 – 44.44%), and ankles (2 – 22.22%). Molecular diagnosis was important to identify family members with intron-22 inversion and to provide appropriate genetic counseling for family planning. A significant statistical difference ($p = 0.019$) was found between carriers and non-carriers in bleeding score. This is an ongoing study and the number of participants needs to be increased in order to draw conclusions. We thank all participants, Fapemig, Fundação Hemominas, and the CGSH of the Ministry of Health.

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QUALITY OF LIFE OF PATIENTS WITH VON WILLEBRAND DISEASE

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von Willebrand disease (VWD) is the most common bleeding disorder due to a qualitative or quantitative disorder of von Willebrand Factor (VWF). There are three main types of the disease (types 1, 2, and 3) and a total of 6 subgroups. Type 1 patients present low levels of VWF; type 2 patients present low activity of VWF, and type 3 patients have a virtual absence of this VWF. VWD is the most common coagulopathy in the human population, but is the least diagnosed. Undiagnosed individuals may present symptoms that are often neglegted. The aim of this study was to analyze the quality of life of patients with VWD who are treated at Fundação Hemominas. Patients were invited to participate in consultations and examinations. The 30 patients included (17 women and 13 men) answered a 36-item brief health questionnaire (SF-36). The analysis of quality of life in women showed that the concept of physical functioning received a median (MED) of 82.5% (IQR 73.8%-95%); the concept of role limitations due to physical health received a MED of 100% (IQR 18.8%-100%); the concept of pain received MED of 61% (IQR 61%-84%); the concept of general health received MED of 77% (IQR 60.8%-88.3%); the concept of energy/fatigue received MED of 57.5% (IQR 33.8%-80%); the concept of social functioning received MED of 81.5% (IQR 25%-100%); the concept of emotional well-being received MED of 83.5% (IQR 33%-100%); and the concept of mental health received MED of 76% (IQR 34%-80%). In men, on the other hand, the analysis of quality of life showed that the concept of physical functioning received a MED of 90% (IQR 70%-100%); concept of role limitations due to physical health received a MED of 25% (IQR 0%-75%); the concept of pain received MED of 72% (IQR 50%-84%); the concept of general health received MED of 67% (IQR 47%-85%); the concept of energy/fatigue received MED of (IQR 50%-80%); the concept of social functioning received MED of 75% (IQR 38%-75%); the concept of emotional well-being received MED of 100% (IQR 67%-100%); and the concept of mental health received MED of 80% (IQR 68%-84%). After analyzing these data, it was not possible to find a significant difference between male and female patients. In conclusion, our study is important to better understand which aspects of quality of life are most impaired in patients with VWD. This study has some limitations because patients were not subdivided by VWD type, and no participants were genotyped yet. This is an ongoing study and the number of participants needs to be increased to draw conclusions. We thank all participants, Fapemig, Fundação Hemominas, and the CGSH of the Ministry of Health.

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RECLASSIFICATION OF PATIENTS WITH HEMOPHILIA A BY MOLECULAR DIAGNOSIS

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The state of Minas Gerais has the third largest population of hemophilia A (HA) patients in Brazil. Fundação Hemominas is the treatment center of reference in the state for clinical and laboratory care of HA patients. Patients are classified according to residual plasma FVIII activity (FVIII:C): Patients with FVIII:C 5%-40% are classified as mild HA; FVIII:C of 1%-5% are classified as moderate HA; and patients with FVIII:C <1% are classified as severe HA. Patients with severe HA often have inversions of introns 1 and 22 in the FVIII gene. The aim of this study was to diagnose inversions of introns 1 and 22 patients with severe and moderate HA. For this purpose, peripheral blood samples were collected from 39 male patients, median (MED) age was 31 years (IQR 19 -42 years),