como os novos agentes de bypassing. Outra nova opção de tratamento que merece destaque é a terapia gênica, que apresenta resultados promissores e um possível potencial curativo da doença, embora ainda não bem definido devido ao risco dos níveis de fator endógeno voltarem a cair. **Conclusão:** Entende-se que a Hemofilia A é uma doença que requer atenção e constante avaliação dos dados epidemiológicos, uma vez que informações de prevalência relativa podem iluminar uma nova visão à abordagem da doença e à compreensão de que sua distribuição nas regiões menos ricas não é menor, menos importante ou menos detectada.

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DISTRIBUIÇÃO EPIDEMIOLÓGICA E LABORATORIAL DA HEMOFILIA EM UM HEMOCENTRO NO NORDESTE DO BRASIL

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Introdução: A hemofilia é uma alteração relativa aos genes, hemorrágica, e de herança recessiva, afetando somente membros do sexo masculino, procedentes de modificações nas unidades fundamental da hereditariedade, codificando os fatores VIII (hemofilia A) ou IX (hemofilia B) da coagulação, localizados no braço longo do cromossomo X contidas no plasma sanguíneo incumbida pela sequência complexa de reações químicas que resultam na formação de um coágulo de fibrina. Toda vez que sucede a ruptura de um vaso, sucede a hemostasia pela ação de várias sequências que irão impedir a perda de sangue que são eles: espasmo vascular, tampão plaquetário, coagulação do sangue e crescimento de tecido fibroso na região do coágulo com o objetivo de vedar o vaso de modo permanente. **Objetivos:** A presente investigação teve como objetivo conhecer a distribuição epidemiológica e laboratorial da hemofilia hereditária e/ou adquirida em um Hemocentro no Nordeste do Brasil. **Metodologia:** Trata-se de uma análise transversal com dados retrospectivos, baseados nas fichas dos indivíduos portadores de coagulopatias, atendidos no ambulatório do Hemocentro, no período de janeiro a dezembro de 2023. Para fins de análise, os dados obtidos foram tabulados, sendo feita análise descritiva. **Resultados:** A pesquisa foi realizada com trinta e quatro (34) pacientes hemofílicos assistidos no ambulatório. Destes todos eram do sexo masculino, sendo que 97,1% (33) são portadores de hemofilia do tipo A e 2,9% (01) hemofilia do tipo B. Quantas localizações 35,3 (12) pacientes residem na grande Aracaju/SE, de Aquidabã e Simão Dias 8,8% (03) indivíduos em cada município. Quanto ao diagnóstico das coagulopatias cerca de 40% apresentou TTPA (Tempo de Tromboplastina Parcialmente ativada) maior que 101s. ultrapassando o valor de referência que é entre 30 a 40s. No que diz respeito ao surgimento de inibidores 0,1% (04) desenvolveram inibidores. Ao ser realizado a incubação do plasma citrado, cerca de 60% (04) amostras indicaram TTPA superior a 101s. **Discussão:** Algumas alternativas terapêuticas foram desenvolvidas para diminuir as complicações que surgem na presença de inibidores, por exemplo, o uso de concentrado de complexo protrombínico (PCCs) ou concentrado de complexo protrombínico ativado (APCCs) podendo assim estimular a formação de um coágulo e cessar a hemorragia, superando o requerimento do FVIII. Porém, este tipo de terapia apresenta algumas limitações, podendo causar o surgimento do inibidor, que irá interferir no processo de coagulação. **Conclusão:** Espera-se que este estudo possa vir produzir impactos positivos para o desenvolvimento de novos estudos, construindo-se para a segurança e qualidade da assistência nas instituições que realizam o diagnóstico e tratamento das coagulopatias hereditárias, e agregar novos conhecimentos aos profissionais de saúde que atuam nessa área.

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PEOPLE WITH MILD HEMOPHILIA ARE AT RISK OF JOINT DISEASE: THE NATIONWIDE COHORT HIN-6 STUDY

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Clinical data on people with mild hemophilia (mPwH) is important to guide their treatment but remains scarce worldwide. In addition, since modern hemophilia therapies can turn severe phenotypes into mild or normal ones, knowing the clinical, therapeutical, and outcomes data of mPwH becomes essential as a proxy of these treatments. We described mPwH who participated in the 6th version of the Hemophilia in the Netherlands (HiN-6) cross-sectional nationwide study. Male mPwH (lowest plasmatic factor activity 5.1%–40.0%) answered a questionnaire about their disease, treatment, and outcomes, in 2019. Questionnaires were elaborated according to age: younger than 12-years (children and

responded by their parents/caregivers), between 12- and 17.9-years (adolescents), and 18-years or older (adults). A total of 387 mPwH responded to the questionnaires. Their median (Interquartile Range; IQR) age was 49.0-years (26.0–62.0), 340 (87.8%) were adults, 29 (7.5%) had past or current inhibitors, 197 (50.9%) had ever been treated with any clotting product, and 12 (3.1%) were currently on prophylaxis. Approximately 74% reported they perceived their disease was not serious or not serious at all, and the different activity groups responded similarly. Recent bleed at any site was reported by 24/47 (51.1%) non-adults, related to the previous quarter, and 96/340 (28.2%) adults, related to the previous year. Joint impairment was reported for 180 joints (22 perceived as severe) by 50/387 (12.9%) mPwH. Age at first hemarthrosis was 10.0 years [5.0–16.0] among 97 (25%) mPwH. Lifetime hospitalization due to joint surgery was reported by 31/358 (8.7%) mPwH. The analyses of PROMIS29 and RAND-26 (quality of life), and HAL (functional abilities) indicated good quality of life and functional abilities. mPwH who participated in the HiN-6 Study reported good social functioning and a non-impactant disease in their lives, although bleeds, joint impairment, and need for joint surgeries still occur.

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ERECTILE DYSFUNCTION IS RELATED TO QUALITY OF LIFE, BUT NOT TO RISK FACTORS FOR CARDIOVASCULAR DISEASES, AMONG ADULT MEN WITH HEMOPHILIA

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Erectile Dysfunction (ED) is a multifactorial disease related to psychosocial determinants and Cardiovascular Disease (CVD). Since Adults with Hemophilia (AwH) may have impaired psychosocial functioning due to the disease and have a high risk of developing CVD, we assumed ED might be prevalent among them. We evaluated perceived ED using a validated self-reported questionnaire in AwH participating in a cross-sectional study to describe CVD and CVD risk factors (HemoCardio Study). Male AwH aged ≥30-years attending a Northeastern Brazilian Hemophilia Treatment Center were invited. Demographic and clinical/therapeutic hemophilia data were collected from medical files. Clinical/therapeutic data about CVD and CVD risk factors were obtained by interview, physical examination, and laboratory tests. Perceived ED was evaluated by the International Index of Erectile Function-Erectile Function Domain (IIEF-6) instrument. Quality of life (QoL) was evaluated by HemoLatin-QoL and EQ-5D. Well-being was evaluated by the Hemophilia Well-Being Index (HWBI). For all questionnaires, higher scores meant better outcomes. A total of 84 AwH was included in the main study. Of the 52 AwH who responded to the IIEF-6, median age was 42.0-years (Interquartile Range [IQR] 36.0–51.8), 43 (83%) had hemophilia A, and 25 (48%) were severe. Median IIFE-6 was 29.0 (IQR 25.3–30.0) and 13 (25%) perceived ED. Perceived ED was associated with severe hemophilia ($p = 0.025$), lower HemoLatin-QoL total score ($p = 0.010$), and lower EQ-5D index ($p = 0.038$), but fairly associated with the use of anti-hypertensive ($p = 0.043$) and antidiabetic ($p = 0.049$). HWBI did not differ between AwH who perceived or did not ED. IIEF-6 had a positive correlation with the emotional functioning domain of the HemoLatin-QoL ($n = 47$, $\varrho = 0.313$, $p = 0.032$), but not with HemoLatin-QoL total score, EQ-5D index, or HWBI. We could not find associations between CVD or CVD risk factors and perceived ED among AwH. However, perceived ED correlated with QoL.

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GENETIC AND NON-GENETIC DETERMINANTS OF SUCCESSFUL IMMUNE TOLERANCE INDUCTION IN PEOPLE WITH SEVERE HEMOPHILIA A

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