SSC Subcommittee Project/Collaborative Project

STANDARDIZATION OF MUSCULOSKELETAL ULTRASOUND IN HEMOPHILIA AND RELATED DISORDERS

Subcommittee

Person responsible (Chair / Principal Investigator): Prof. Roberta Gualtierotti

Description Abstract

State the application's broad, long-term objectives and specific aims, making reference to the health relatedness of the project. Suggested length is 2-3 paragraphs.

The advent of novel effective treatment and the identification of the need to achieve a higher trough level for patients with hemophilia has changed the landscape of the management of these patients, allowing to change the target from survival and prevention of life-threatening complications to prevention of musculoskeletal complications and improvement of quality of life.

Unfortunately, the subjective report by the patient and the physical examination by the physician are not sufficiently reliable to identify joint bleeding, whereas ultrasound has emerged as a more sensitive non-invasive tool for the differential diagnosis of acute joint pain and early recognition of damage [1].

However, a lack of standardisation in the definition of the pathological changes may lead to confusion and an incorrect evaluation of the presence and progression of joint damage, subsequently delaying an adequate pharmacological and non-pharmacological personalised management when a window of opportunity is still open [1, 2].

We here propose a process of standardization of the definitions of the articular complications associated with hemophilia to improve management of patients.

Design and methodology (Data expected to collect, sample size and statistical analysis):

Describe concisely the research design and methods for achieving these goals. Suggested length 2-3 paragraphs

The first part of the project (first 12 months) will be aimed at collecting information on the current application of musculoskeletal ultrasound in patients with hemophilia and related disorders and a consensus on the definitions of the main ultrasound biomarkers of hemophilic arthropathy: joint bleeding, synovitis and osteochondral damage.

The second part (24-36 months) will be aimed at validating the so defined ultrasound biomarkers in a large population by correlating joint bleeding with self-reported annual bleeding rates and pain and the findings of joint damage with FVIII levels and other confounding (e.g. BMI, age, sex or gender).

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Study population (Inclusion, exclusion, eligibility) (patient population; recruitment of participating institutions/physicians and subjects; minimum number needed; expected number):

Suggested length 2-3 paragraphs

For the first part of the project, we will send a questionnaire to the main Comprehensive care centers for the care of hemophilia and related disorders with proven experience in musculoskeletal ultrasound evaluation worldwide. For the consensus, we will involve hematologists, internists and rheumatologists expert in musculoskeletal ultrasound evaluation of patients with hemophilia and related disorders.

For the second part of the project, we will involve a population of patients with hemophilia of all degrees of severity and Von Willebrand Disease (VWD) of any type of severity of any age, sex and gender. Considering the prevalence of hemophilia and VWD we expect a population of at least 1000 patients with hemophilia and 2000 patients with VWD

Expected timeline:

Project stage/set up Jan 2024
Launch June 2024
Finalization/analysis Jan 2027
Reporting May 2027
Duration 36 months

Expected outcomes (ie. publications):

1. Survey on musculoskeletal ultrasound (Guidance document)
2. Consensus paper on shared definitions (SSC Communication)
3. Validation of the ultrasound biomarkers of musculoskeletal ultrasound (Original article)

Description of project set/up and management, needed infrastructure and resources (summary):

The Centers and hematologists, internists and rheumatologists that will be involved will need to demonstrate their experience in musculoskeletal ultrasound assessment of hemophilic arthropathy (possible collaborations: Prof. Matteo Dario Di Minno, Prof. Suchitra Shridhar Achayra, Dr. Samantha Gouw, Prof. Alok Srivastava, etc). We plan to involve a methodologist in the design of the study (e.g. Prof. Frits Rosendaal)

Possible references:

1. Bakeer N et al. Musculoskeletal ultrasound in hemophilia: results and recommendations from a global survey and consensus meeting. Res Pract Thromb Haemost. 2021;5(5):e12531
2. Lewandowska M, Nasr S, Shapiro AD. Therapeutic and technological advancements in haemophilia care: Quantum leaps forward. Haemophilia. 2022 May;28 Suppl 4:77-92. doi: 10.1111/hae.14531. PMID: 35521732