2.01 Hemophilia - clinical and treatment issues

FUSION 2: VALIDATION AND CLINICAL PERFORMANCE OF AN INTEGRATED ULTRASOUND-FUNCTIONAL ASSESSMENT SYSTEM IN HEMOPHILIA

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Background and Aims. FUSION (Functional & Ultrasound Integrated Observation in Hemophilia) is an integrated musculoskeletal assessment framework combining point-of-care ultrasound (HEAD-US) with a simplified functional evaluation. While ultrasound and clinical joint scores provide complementary information, the extent to which functional performance contributes independent information beyond established tools remains unclear. This study aimed to validate the FUSION composite score and investigate whether its functional component captures clinically relevant dimensions not explained by HEAD-US or Hemophilia Joint Health Score (HJHS) 2.1 alone.

Materials and Methods. We conducted a prospective multicenter cross-sectional study including 121 patients with hemophilia A, hemophilia B, or von Willebrand disease undergoing integrated assessment with HEAD-US, HJHS 2.1, and FUSION. The FUSION score combines HEAD-US evaluation of elbows, knees, and ankles with a weighted simplified functional score (F×4) derived from six standardized motor tasks. Convergent validity was assessed using Spearman correlation and Steiger's z-test. Variance decomposition and residual analyses were performed to identify the proportion and nature of information uniquely captured by FUSION.

Results. FUSION showed strong convergent validity with HJHS(ρ=0.764, $p<0.001$), comparable to the correlation observed between HEAD-US and HJHS (ρ=0.776, $p<0.001$; Steiger p=0.82). However, addition of the functional component significantly increased explained variance beyond HEAD-US alone (ΔR^2=0.070, $p<0.001$), supporting an independent contribution of functional assessment. Variance decomposition demonstrated that 25.5% of the variance captured by FUSION was not explained by HJHS. Notably, 43.1% of the variance of the functional component (F×4) remained unexplained by HJHS, indicating that FUSION captures a substantial non-overlapping functional domain. Discordance analyses further supported the complementary role of FUSION. Among patients with HJHS=0, FUSION identified abnormalities in 53.8% of cases. In patients with HEAD-US=0, FUSION detected abnormalities in 55.0% versus 30.0% with HJHS alone (p=0.013), with all additional detections driven by the functional component.

Conclusions. FUSION preserves strong alignment with established hemophilia joint assessment tools while simultaneously identifying a substantial and clinically relevant functional domain not captured by HJHS or structural ultrasound evaluation alone. These findings support the concept of a multidimensional structural-functional assessment model and suggest that current joint-focused scores may underestimate global biomechanical and compensatory impairment in hemophilia.