

Abstract

Joint Hypermobility Syndrome (JHS) is considered virtually indistinguishable from Ehlers Danlos Syndrome Hypermobile Type (EDS-HT), although full consensus may not occur until the genetic marker is found. Although this rare, heritable connective tissue disorder (HCTD) has a variety of potentially disabling systemic effects, clinical scepticism of its impact on the patient still exists. The myriad of associated dysfunctions affect virtually every body system. The financial burden of the syndrome is likely to be considerable.

Recently, strategies for recognition and management of rare diseases have been developed at international and national level. Information technology can play a key role in the implementation of these strategies.

Education, communication and information gaps exist at almost every stage of the EDS-HT patient journey. Ideally, all clinicians involved in the care of EDS-HT patients would have timely access to valid, reliable and complete patient data at point of care.

Standardisation of the essential information or data that is relevant to the care of EDS-HT patients, can ensure that data is complete, relevant comparable and capable of aggregation for population studies, thus attracting interest and funding for further research. This research presents the methodology and the first iteration for development of an EDS-HT minimum dataset.

The design and development of appropriate informatics solutions may facilitate safe storage and timely retrieval of data and an opportunity to support and transform care for patients with EDS-HT.