Inpatient Rehabilitation for Ehlers-Danlos Syndrome
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Background
Ehlers-Danlos syndrome (EDS) is a heritable connective tissue disorder caused by a mutation in the COL5A1 and 2 genes which encode the alpha 1 and 2 chains of type V collagen. This functionally defective collagen protein characterizes EDS Types I and II and causes a continuum of clinical findings listed below, most notably skin hypertextensibility, tissue fragility, and generalized joint hypermobility. Most patients with EDS have the hypermobility type (type III), which manifests as generalized severe joint laxity with dislocations and chronic pain. These manifestations naturally lead to functional impairment, but have rarely been described in the rehabilitation literature.

Major Diagnostic Criteria
- Skin hyperextensibility
- Wideden atrophic scarring
- Joint hypermobility (both large and small joints, from mild to severe)
- Positive family history

Minor Diagnostic Criteria
- Smooth, velvety skin
- Molluscoid pseudotumors
- Subcutaneous spheroids
- Complications of joint hypermobility
- Muscle hypotonia
- Easy bruiting
- Manifestations of tissue extensibility (hiatal hernia, pelvic viscera prolapse)
- Surgical complications (hernias)

Case Description
A 22-year-old female with a recent diagnosis of EDS was admitted to acute inpatient rehabilitation after 9 months of immobility due to multiple ligamentous injuries of her ankles requiring surgical repairs. Prior to her ankle surgeries she had been using crutches for ambulation due to severe ankle instability, but after her period of bedrest following surgery, she required a power wheelchair and assistance with most ADLs and even static sitting. She had made little progress in outpatient physical and pool therapy. She was on an opioid regimen for chronic pain management, and was also coping with anxiety and depression which had begun over the last year. She presented with severe deconditioning, chronic pain, and a marked functional decline, requiring maximum to contact guard assistance for mobility and most functional activity.

Examples of isometric stabilizing exercises used. From left: Hip flexion with 2 lb weight, external arm rotation (top), knee flexion against pillow (bottom), internal arm rotation, neck lateral flexion and rotation (top), neck flexion (bottom).

Several different orthoses were created and used during her inpatient stay. McConnell (rigid) taping of the patellae, as well as wrist braces were used to prevent dislocation. As she was unable to use her custom solid AFOs due to hip and SI joint dislocation from weight of material, elastic bandages and air casts were used to maintain neutral ankle position and medio-lateral support. The orthotics department created a custom shoulder orthosis described in detail at right.

She showed improvement during her stay; with strengthening and education, her bed mobility and transfers required less time and caused less pain and fewer dislocations. After 2 weeks she returned home with her family to continue in outpatient therapies.

Discussion
A multidisciplinary rehabilitation approach was used to address this patient’s profound deconditioning and maximize functional outcomes given the limitations of her severe hypermobility. The rehabilitation medicine team managed multiple joint dislocations (including reducing shoulder dislocations several times). Her regimen of opioid medications required adjustment and monitoring given the variability in her pain level resulting from increased activity and dislocations upon starting therapies. Psychiatry and psychology services were consulted for input in managing her anxiety, depression, and disordered sleep which had worsened with the decline in her functional status. Initially she was limited to low repetitions of isometric exercises at all joints, but she progressed to active range of motion exercises with increased repetitions. Core strengthening exercises were incorporated into active range exercises. Therapists also focused on education on proper joint alignment, abdominal bracing and setting of proximal musculature during functional mobility. Basic modalities including hot and cold packs and relaxation also helped with her joint pain.

Shoulder Orthosis
The design of this left shoulder orthosis aims to prevent the disruption of the synergistic action of the multiple joints within the shoulder (scapulohumeral rhythm), which alters the center of rotation of the humeral head and allows excessive excursion from the glenoid fossa. The orthosis includes a flexible inner cap which moves with the scapula and clavicle through the scapulohumeral rhythm, as well as providing an attachment point for the control strap proximal and medial to the glenohumeral joint to assist with joint congruency. The neoprone shoulder support maintains the custom cap in place throughout the shoulder range of motion.

The custom shoulder orthosis. From left: The molded inner cap, the orthosis in place.

Conclusion
The manifestations of EDS can cause pain and functional decline, complications of immobility, and psychosocial implications. There are few reports in the literature of the benefits of acute inpatient rehabilitation for these complex patients. Our patient benefitted from a multidisciplinary approach, with contributions from physical and occupational therapy, psychology, orthotics, and physiatry. Acute inpatient rehabilitation is an appropriate setting to address the many features of this disorder and promote safe functional activity for these complex patients.

References