Jessica Rolle

**Multidirectional Instability and Ehlers-Danlos Syndrome:**

**Evaluation and Treatment of the Shoulder**

*ANATOMY AND PHYSIOLOGY*

The shoulder complex is comprised of four joints, including the glenohumeral, acromioclavicular, sternoclavicular, and scapulothoracic joints. However, for the purposes of this project, the main focus will be on the glenohumeral (GH) joint. There are numerous static and dynamic stabilizers of the shoulder, which provide support and allow for proper movement patterns of the upper extremity. The static stabilizers are non-contractile units such as the joint capsule, glenoid labrum, and ligaments. The joint capsule surrounds the GH joint and gives multidirectional stability for the joint while the glenoid labrum increases the surface area of the glenoid. The superior glenohumeral ligament has attachment sites on the supraglenoid tubercle of the scapula and the proximal tip of the lesser tubercle of the humerus; therefore, it helps to resist inferior motion of the humerus.1 The middle GH ligament has attachment sites on the supraglenoid tubercle, anterosuperior region of the labrum, and humeral head, providing stability when the humerus moves anteriorly in relation to the glenoid and when the humerus is externally rotated.1 The inferior GH ligament (IGHL) is comprised of an anterior and posterior band, as well as an axillary pouch between the two bands.2 This IGHL complex acts as a sling and can resist either anteroinferior motion or posteroinferior motion depending on arm position.2 Finally, the coracohumeral ligament attaches proximally to the dorsolateral base of the coracoid process and distally to the greater tuberosity of the humerus.1 Due to the anterosuperior location of this ligament, it can resist posterior and inferior motion.

Dynamic stabilizers of the shoulder consist of contractile units, or muscles. The rotator cuff, including supraspinatus, infraspinatus, teres minor, and subscapularis, is a critical group of muscles that work together to provide shoulder stability and ensure that the humeral head moves appropriately within the joint during shoulder movement.3 Numerous other muscles surround the shoulder joint and help to provide additional support, such as the biceps, triceps, pectorals, deltoid, trapezius, teres major, serratus anterior, rhomboids, and latissimus dorsi.

Despite all of these static and dynamic stabilizers, the glenohumeral joint is the least stable joint in the body. The humeral head has 3-4 times greater surface area than the glenoid, which can be likened to a golf ball on a tee.4 There is an inverse relationship between mobility and stability; the GH joint allows significant mobility in all planes, but it lacks stability. When considering arm elevation, it is important to consider the contributions of the glenohumeral joint and the scapulothoracic (ST) joint. From 0-30 degrees of elevation, the glenohumeral joint contributes all of the motion.4 However, from 30-180 degrees, two thirds of the motion comes from the GH joint and one third comes from the ST joint.4 Scapulohumeral rhythm can be altered when the shoulder stabilizing structures are injured or weak.

*SHOULDER INSTABILITY*

Shoulder instability can have either traumatic or atraumatic etiologies. Traumatic shoulder dislocations and subluxations typically (95%) occur in individuals who fall on an outstretched hand and incur an abduction and ER force.4 This leads to an anteroinferior dislocation of the humeral head from the glenoid and damage to the middle GH ligament and anterior band of the inferior GH ligament. The other 5% of individuals who have instability of traumatic origin sustain a posterior dislocation or subluxation due to an adduction and internal rotation force.4

Atraumatic shoulder instability is caused by either genetics or an underlying condition affecting the individual’s ligaments. Multidirectional instability (MDI) and Ehlers-Danlos syndrome (EDS) are two such examples and will be the primary focuses of this review.

*MULTIDIRECTIONAL INSTABILITY*

Multidirectional instability (MDI) can be defined as having ligamentous laxity of more than one ligament surrounding the GH joint, which results in increased arthrokinematic motion in multiple directions. MDI is not likely the primary diagnosis that brings a patient to physical therapy, however. Patients are more likely to have sustained an injury, such as a shoulder dislocation with resulting pain, decreased mobility, and decreased functional use. The shoulder instability special tests that can be utilized during the initial examination are the: sulcus, rockwood, anterior load and shift, apprehension/relocation, push-pull, and jerk tests.4 Since patients with MDI have instability in all directions, they may have positive results on multiple special tests.

*SURGICAL OPTIONS FOR MDI*

Conservative treatment should always be implemented initially for patients with MDI. While traumatic shoulder injuries are more likely to require surgery, patients with atraumatic shoulder instability tend to have better outcomes with non-surgical approaches. However, some patients with multidirectional instability do end up requiring surgery as a last resort if non-surgical treatment is unsuccessful. The three main surgical options for MDI are thermal shrinkage, capsular shift/plication, and rotator interval closure.5-7 The idea behind each of these surgical procedures is to shrink the joint capsule and tighten the structures within the rotator interval.5-7 Each of these techniques has only proven to have satisfactory results, with a thermal shrinkage failure rate of 31-47%.5,6 Patients then must go through rehabilitation to recover from these procedures, which could set them back for a longer period of time.

*EHLERS-DANLOS SYNDROME*

 Ehlers-Danlos syndrome (EDS) is a connective tissue disorder where the individual has a genetic collagen deficiency. The typical characteristics across all six EDS subtypes are skin extensibility, tissue fragility, and joint hypermobility due to ligamentous laxity.8 EDS occurs in about 1 in 2,500 to 1 in 5,000 individuals worldwide.8 However, it is suspected that EDS is widely under diagnosed and most individuals go many years before an official diagnosis is confirmed.9-11 EDS is inherited and the specific subtype of EDS runs true in families.

The hypermobility type of EDS is the most common of the six subtypes. The typical characteristics of this type of EDS are joint hypermobility, velvety skin with or without extensibility, and chronic pain.8 Physical therapists will be treating patients with EDS-hypermobility type for dislocations, subluxations, sprains, pes planus, and/or postural abnormalities. Patients with classical EDS have more skin involvement, with hyperextensibility, scarring, and bruising, as well as joint hypermobility.8 These individuals may also seek physical therapy for joint dislocations or subluxations. The vascular subtype of EDS is the most serious type and can be life threatening if not managed appropriately. These patients are at risk for arterial or organ rupture due to the fragility of their arteries, intestines, uterus, and skin.8 Kyphoscoliosis is a rare subtype of EDS and is characterized by joint laxity, muscle hypotonia, scoliosis, and tissue and eye fragility.8 These patients are more likely to be diagnosed early in life and require physical therapy for gross motor development, ambulation, and postural education. Unfortunately, many of these patients will lose their ability to ambulate by 30 years old.8 The fifth subtype of EDS is arthrochalasia. Patients with this type have congenital hip dislocations seen in early childhood, joint hypermobility, and skin involvement.8 Finally, dermatosparaxis EDS is another very rare subtype. Characteristics of this include skin fragility, easy bruising, sagging skin, and large hernias.8 While the hypermobility, classical, vascular, and arthrochalasia types of EDS are autosomal dominant, the kyphoscoliosis and dermatosparaxis types are autosomal recessive.8 Genetic testing can be performed for each of the subtypes except the hypermobility type, which has no known genetic marker.8 Diagnosis is made purely by the patient’s clinical presentation and family history.

Differential diagnosis is an important evaluation component for physical therapy. Marfan syndrome is a similar connective tissue disorder that primarily affects the heart, blood vessels, eyes, bones, and lungs.12,13 Cutis laxa is characterized by extensive skin involvement including loose, sagging skin.14 Loeys-Dietz syndrome mimics the vascular subtype of EDS and these patients are also at risk for aneurysms.12 Finally, patients with Williams syndrome typically have joint laxity, low muscle tone, and connective tissue abnormalities.12 Each of these conditions can be diagnosed by genetic testing.

The Beighton scale is frequently utilized in physical therapy practice to determine whether an individual has generalized hypermobility. It can therefore be one of several tools to use for patients with EDS. The patient is scored based on their ability to: 1) passively dorsiflex the fifth metacarpophalangeal joint to >90 degrees (test each hand), 2) oppose the thumb to the volar aspect of the ipsilateral forearm (test each hand), 3) hyperextend the elbow to >10 degrees (test each elbow), 5) hyperextend the knee to >10 degrees (test each knee), and 5) place hands flat on the floor without bending the knees.15 A score of 5/9 or greater is considered positive for hypermobility.15

*PHYSICAL THERAPY INTERVENTIONS FOR MDI AND EDS*

The most important component of the treatment plan for patients with multidirectional instability or Ehlers-Danlos syndrome is neuromuscular re-education. Research has shown that individuals with inherent joint hypermobility tend to have impaired proprioception and kinesthetic awareness.8,16 Joint receptors have a reduced capability to detect subtle changes in body position; therefore, the patient may not recognize when the joint is moving too far and this will increase their injury risk. Although these patients have impaired proprioception, the evidence shows that exercise interventions focusing on neuromuscular control and joint stabilization can make statistically and clinically significant improvements in proprioception.16,17 These control and stability exercises should be performed at various positions throughout the patient’s full (including hypermobile) range.18 In everyday activities, the patient will be moving into this hypermobile range (intentionally or not), so it is critical to have control within the whole range of motion.

As mentioned previously, there are both static and dynamic stabilizers. Throughout the physical therapy intervention, it would be critical to target the dynamic shoulder stabilizers when implementing a strengthening program. These muscles must be strong to compensate for the weak, loose ligaments and capsule surrounding the joint. It is recommended to perform exercises with low resistance and high repetitions in order to increase the patient’s resting muscle tone and overall joint stability.19 Research has shown that closed kinetic chain exercises may be more effective in improving proprioception and balance in patients with joint hypermobility.17,20

Restoring appropriate scapular motion is another key component of physical therapy treatment for this population of patients with MDI or EDS. In order to complete functional tasks, the shoulder complex must have full, painfree motion. It is important, therefore, to examine the soft tissues and muscles surrounding the joint to determine if there are any significant restrictions.20 Manual therapy can be utilized to treat trigger points or muscle spasms that are caused by muscle guarding and altered movements patterns due to pain.

Patient education is an essential factor when treating a patient with MDI or EDS. Postural awareness is one example; if the patient has rounded shoulders with protracted scapulae, the glenoid will be pushed forward and will place the shoulder in a more vulnerable, unstable position. Patients should also avoid stretching into their joints’ hypermobile ranges since this will place added stress on the joint structures. Although many of these patients will automatically avoid high-impact activities, therapists should encourage patients to avoid putting their unstable joints at risk.12,19 Patients with MDI and EDS should perform their home exercise programs daily and understand that their condition will need to be managed throughout their lives.

There are several outcome measures that can be used with patients with MDI or EDS. In particular, the Melbourne Instability Shoulder Scale and Western Ontario Shoulder Instability Index have been shown to be valid and specific to instability.21 Other more general shoulder outcome measures include the Simple Shoulder Test, the Shoulder Rating Questionnaire, and the Disabilities of the Arm, Shoulder, and Hand Questionnaire. These indices help to establish a baseline for these patients in terms of function, pain, and health-related quality of life, as well as to document changes over time.21

*OTHER CONSIDERATIONS FOR EDS*

Some patients with EDS may benefit from bracing, assistive devices, or orthotics in order to provide some external support for their lax joints.12,22 Utilizing taping or strapping techniques may offer some tactile cueing to stimulate mechanoreceptors and proprioceptors in the soft tissues. Physical therapists should be cautious when working with patients with EDS, as they have a tendency to bruise easily and may have chronic pain. Postural orthostatic tachycardia syndrome is another common comorbidity with EDS and may affect a patient when changing position too quickly.8 Joint manipulation and mechanical lumbar and cervical traction should be avoided since these will place unnecessary stress on the joints.

**References**

1. Burkart AC, Debski RE. Anatomy and function of the glenohumeral ligaments in anterior shoulder instability. *Clin Orthop Relat Res*. 2002;400:32-39.

2. O’Brien SJ, Neves MC, Arnoczky SP, et al. The anatomy and histology of the inferior glenohumeral ligament complex of the shoulder. *Am J Sports Med*. 1990;18(5):449-456.

3. Hughes M. Glenohumeral joint anatomy, stabilizer, and biomechanics. *Orthobullets*. <http://www.orthobullets.com/sports/3032/glenohumeral-joint-anatomy-stabilizer-and-biomechanics>. Updated 01/04/14. Accessed 01/20/14.

4. Magee DJ. *Orthopedic Physical Assessment*. 5th ed. St. Louis, MO: Saunders Elsevier; 2008.

5. Toth AP, Warren RF, Petrigliano FA, et al. Thermal shrinkage for shoulder instability. *HSS J*. 2011;7(2):108-114.

6. Bell JE. Arthroscopic management of multidirectional instability. *Orthop Clin North Am*. 2010;41(3):357-365.

7. Provencher MT, Mologne TS, Hongo M, Zhao K, Tasto JP, An KN. Arthroscopic versus open rotator interval closure: biomechanical evaluation of stability and motion*. Arthroscopy*. 2007;23(6):583-592.

8. The Ehlers-Danlos National Foundation. <http://www.ednf.org>. Updated 2014. Accessed Sep 2013-Feb 2014.

9. Castori M. Ehlers-Danlos syndrome, hypermobility type: an underdiagnosed hereditary connective tissue disorder with mucocutaneous, articular, and systemic manifestations. *ISRN Dermatol*. 2012;2012:751768.

10. Adib N, Davies K, Grahame R, Woo P, Murray KJ. Joint hypermobility syndrome in childhood. A not so benign multisystem disorder? *Rheumatology*. 2005;44(6):744-750.

11. Grahame R. A Rheumatologist’s Perspective Over Four Decades [PowerPoint]. Summer 2011. Baltimore, Maryland: Ehlers-Danlos National Foundation Learning Conference.

12. Levy HP. Ehlers-danlos syndrome, hypermobility type. *Gene Reviews*. <http://www.ncbi.nlm.nih.gov/books/NBK1279/>. Updated 2012. Accessed 01/15/14.

13. What Are the Signs and Symptoms of Marfan Syndrome? National Institutes of Health. <http://www.nhlbi.nih.gov/health/health-topics/topics/mar/signs.html>. Updated 10/01/10. Accessed 01/21/14.

14. Cutis Laxa. Genetics Home Reference. <http://ghr.nlm.nih.gov/condition/cutis-laxa>. Updated 06/2014. Accessed 01/21/14.

15. Sanches SHB, Osorio FL, Udina M, Martin-Santos R, Crippa JAS. Anxiety and joint hypermobility association: a systematic review. *Rev Bras Psiquiatr*. 2012;34(Suppl1):553-568.

16. Sahin N, Baskent A, Cakmak A, Salli A, Ugurlu H, Berker E. Evaluation of knee proprioception and effects of proprioception exercise in patients with benign joint hypermobility syndrome. *Rheumatol Int*. 2008;28(10):995-1000.

17. Ferrell WR, Tennant N, Sturrock RD. Amelioration of symptoms by enhancement of proprioception in patients with joint hypermobility syndrome. *Arthritis Rheum*. 2004;50(10):3323-3328.

18. Pacey V, Tofts L, Adams RD, Munns CF, Nicholson LL. Exercise in children with joint hypermobility syndrome and knee pain: a randomised controlled trial comparing exercise into hypermobile versus neutral knee extension. *Pediatr Rheumatol Online J*. 2013;11(1):30-41.

19. Ehlers-Danlos Syndrome. Physiopedia. <http://www.physio-pedia.com/Ehlers-Danlos_Syndrome>. Accessed 01/26/14.

20. Wilk KE, Macrina LC, Reinold MM. Non-operative rehabilitation for traumatic and atraumatic glenohumeral instability. *N Am J Sports Phys Ther*. 2006;1(1):16-31.

21. Plancher KD, Lipnick SL. Analysis of evidence-based medicine for shoulder instability. *Arthroscopy*. 2009;25(8):897-908.

22. Management of Ehlers-Danlos Syndrome. EDSers United. <https://www.edsers.com/management.html>. Accessed 01/26/14.