If you, or a family member, have been informed that you have genetic-based rotator cuff disease (GBRCD), here is information that may help explain the diagnosis.

**Why:**
GBRCD is primarily structural or anatomical in nature, arising from a too-narrow subacromial space which is located just superior (above) the rotator cuff (RC). Due to this “impingement”, the outer-surface of the RC will be subjected to increase wear, thereby increasing the risk for a tear at an early age (less than 45 years of age).

Anatomically, the subacromial space is formed by the acromion above and the humeral head below two of the RC tendons. The coracoacromial ligament (CAL) is typically one of the main contributors to the pathology. This ligament originates at the coracoid and inserts on the acromion. The insertion typically ends at the tip of the acromion. However, in GBRCD, the CAL is too big, either too long and/or too thick, thus decreasing the space the rotator cuff has to work efficiently and in a healthy manner. Another contributing factor is the shape of the acromion. In GBRCD, the acromion most often is not linear/straight; it may be curved or hooked, contributing to decreased space and increased likelihood of formation of a bony projection at insertion of CAL (enthesophyte).

**Diagnosis:**
Pain is what typically brings symptomatic GBRCD patients to clinic. Treatment will be based upon their signs and symptoms, including but not limited to pain, weakness, decreased function, and limitations in range of motion. Treatment options are outlined below. If someone is at an increased risk to develop RC pathology due to genetics (typically transmitted via same genders with up to a 25% risk for same gender, less than 10% for opposite gender), careful monitoring is the primary course of treatment.

However, if a non-symptomatic patient incurs an injury that does cause pain, weakness and decreased function, they should be seen immediately. If you delay treatment in an injured shoulder or a shoulder with decreased function with known GBRCD, you are putting yourself at an increased risk of having a non-repairable rotator cuff tear.
Signs and Symptoms

- Recurrent, constant pain, particularly with overhead activities
- Pain at night that prevents you from sleeping on the affected side
- Muscle weakness, especially when attempting to lift the arm
- Catching, grating or cracking sounds when the arm is moved
- Limited motion
- May be triggered by a specific incident, specifically falling on hand or elbow, pulling mechanism

Risk Factors

- Family history of genetic based rotator cuff disease with/without rotator cuff tear
- Repetitive overhead motion, such as painting a ceiling, pitching, or carpentry
- Heavy lifting
- Excessive force, such as a fall
- Degeneration due to aging, including a reduction in the blood supply to the tendon

Treatment Options

Upon examination, you will have your range of motion and strength tested, as well as have the top and back of your shoulder examined to see if the muscles have begun to shrink (atrophy). At that time, X-rays may be taken to help the doctor see any problems with the bones. Another imaging test, MRI (magnetic resonance imaging), may be required to confirm a rotator cuff tear.

Conservative Treatment

In most cases, initial treatment for known GBRCD is non-surgical. Treatment involves a combination of modalities including:

- Activity modification
- Physical therapy to assist with strengthening the RC to decrease the impingement

Treatment with known rotator cuff tear with GBRCD:

- Rest. If the tear is due in part to overuse, resting the shoulder may help combined with activity modification
- Physical therapy to restore flexibility and strengthen the shoulder muscles.
- Corticosteroid injections can help reduce pain, but cannot be repeated frequently because they can also weaken the tendon (usually no more than two or three injections are given).
- Nonsteroidal anti-inflammatory medications may help control pain.

Surgical Intervention

There are several surgical options to treat rotator cuff tears, depending on the size, depth, and location of the tear. If other problems with the shoulder are discovered, they will be corrected as well. Structures and conditions in the shoulder that often are involved with the development of a rotator cuff tear include acromioclavicular joint (AC joint) arthritis, biceps tendon degeneration, and coracoid impingement.

- Arthroscopy- small instruments are inserted into small incisions, and are used to remove bone spurs or inflammatory portions of muscle and to repair small tears.
- A mini-open repair combines arthroscopy and the use of small incisions to treat full-thickness tears.
- Open surgery is required to repair the rotator cuff tear that is medium to large in size.

♦ Materials borrowed from the American Academy of Orthopaedic Surgeon (more information can be found at www.aaos.org)
And from Shoulder Doc (more information can be found at http://www.shoulderdoc.co.uk/index.asp)