

Cutis laxa is a rare disorder that affects males and females in equal numbers. The disorder has been reported in approximately 400 families worldwide. Cutis laxa is estimated to affect 1 in 1,000,000 individuals in the general population. However, because cases may go misdiagnosed or undiagnosed determining the true frequency of cutis laxa in the general population is difficult. Cutis laxa affects individuals of all races and every ethnic group. A diagnosis of cutis laxa is based upon identification of characteristic symptoms, a detailed patient history, a thorough clinical evaluation and a variety of specialized tests. Surgical removal and microscopic examination (biopsy) of affected skin can reveal characteristic changes in elastic fibers. Distinguishing between the specific genetic forms of cutis laxa can be difficult. Molecular genetic testing can confirm a diagnosis of an inherited form cutis laxa and establish the specific, underlying subtype in some cases. Molecular genetic testing can detect mutations in specific genes known to cause cutis laxa, but is available only as a diagnostic service at specialized laboratories.