

Who are Geneticists and Genetic Counselors?

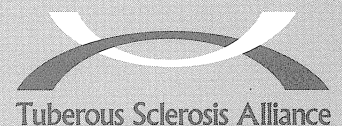
Hopefully, one of the first things you heard when your child or family member was diagnosed with tuberous sclerosis complex (TSC) was "you need a referral to see a geneticist" because TSC is a genetic condition. Before your child's diagnosis of TSC you may have never heard of a geneticist. Who are geneticists and what do they do? There are several different types of geneticists: M.D. Medical Geneticists, Ph.D. Medical Geneticists and Master's level Genetic Counselors comprise the three main groups of geneticists in clinical practice.

An M.D. Medical Geneticist is a physician who specializes in the clinical practice of medical genetics. To specialize in medical genetics, a physician must undergo an examination process that ensures their skills in practicing genetic medicine (see below for a description of the certification process). There are currently slightly more than 1,000 physicians who are board-certified to practice clinical medical genetics in the United States. Among Medical Geneticists, the majority (>60%) are also board-certified in Pediatrics. All Medical Geneticists have completed medical school and most complete two additional training periods, a three- or four-year period (residency) in one area of specialty (i.e. Pediatrics, Internal Medicine, Obstetrics & Gynecology) as well as a minimum of a two-year training period specifically in genetics (referred to as a residency or fellowship).

Ph.D. Medical Geneticists are health care professionals who specialize in clinical genetics. While these doctors are not physicians, they are trained in the diagnosis and care of individuals with genetic conditions. Ph.D. level geneticists must also undergo the board certification examination process described below. There are approximately 150 board-certified Ph.D. Medical Geneticists in the United States. These individuals have usually obtained their terminal degree (Ph.D., requiring approximately five to seven years of study) in one area of genetics and then further trained for approximately two years in the field of medical genetics. In most instances, Ph.D. Medical Geneticists work with M.D. Medical Geneticists in clinics or other diagnostic settings.

M.D. and Ph.D. Medical Geneticists are certified by the American Board of Medical Genetics (ABMG). The board examination for Medical Genetics began in the early 1980s and in the 1990s the American Board of Medical Specialties recognized Medical Genetics as a Medical Specialty. The examination process includes: an education in medical genetics at a recognized institution that has been "accredited" or recognized as a genetics training program, documented care of at least 150 patients and families affected with genetic conditions and completion with a passing grade of a comprehensive examination. These examinations have been given every three years; however, the cycle is being changed to every two years as of 2003. After certification, Medical Geneticists are required to keep abreast of the field via continuing education.

The last category of geneticists is Genetic Counselors. Genetic Counselors are Master's level-trained health care professionals educated in conveying genetic risk and diagnostic information to patients. Genetic Counselors are board-certified by the American Board of Genetic Counseling via an examination process that is similar to that undergone



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by the M.D. and Ph.D. medical geneticists. There are more than 1,500 board-certified genetic counselors in the United States. Genetic Counselors usually obtain a specialized Masters of Science from a recognized training program that is about two years in duration. Genetic Counselors work in a number of sites and settings, including prenatal diagnosis, pediatric clinics, hereditary cancer counseling and public health administration.

Most referrals to genetics clinics are babies born with birth defects or children who have developmental delay and/or unusual physical features. Medical Geneticists are the doctors' doctors. Most referrals to Medical Geneticists come from other doctors sending their patients to gain additional information. The geneticist, along with their team of genetics professionals, tries to meet three goals in their association with the patient: diagnosis, prognosis and recurrence risk. Making a diagnosis (giving a name to the condition) leads to information about the prognosis (what is going to happen in the future) for the affected patient. Making a diagnosis enables the geneticist to determine a recurrence risk for the family. A recurrence risk simply means the risk (or odds) that a subsequent child will be affected with the same condition. The parents of the affected child are usually the most concerned about recurrence risk; however, other family members (for example, aunts or uncles of the affected child) often desire the information as well.

In the case of diagnosing TSC, the genetic professional's job is to convey to the individual or parent the genetic facts about TSC. TSC is a genetic disease that is caused by a gene that is changed (mutated) in some way, which can then be passed onto future generations. However, it is important to remember that approximately two-thirds of the time when a child is diagnosed with TSC, neither of the parents has TSC. We know that in TSC it is very common for the gene to be changed before or during the formation of a new individual. This change in the genetic material is referred to as a new (or sporadic) mutation. In these families, a medical geneticist or other physician familiar with TSC can examine the parents and document that they do not have signs of TSC, making a new mutation in the child the most likely reason for TSC. In the other one-third of cases, there are one or more family members who have TSC, leading to additional issues for the geneticist to discuss. The genetics professional helps in coordinating studies of other family members in order that they obtain needed care. The most recent important duty of the genetic professional is to discuss with the family options for DNA testing. DNA testing determines the exact change, or mutation, that has occurred in one of the two TSC-causing genes (TSC1 or TSC2). A gene produces a protein necessary for the functioning of the human body. In persons with TSC, the mutation in one of the two TSC genes causes the signs of symptoms of the condition. If the mutation that caused TSC in an individual or family can be detected, further testing can then be offered to more easily and precisely diagnose possibly affected family members or for prenatal diagnosis of TSC.

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***Tuberous Sclerosis Alliance Information Sheets are intended to provide basic information about TS. They are not intended to, nor do they, constitute medical or other advice. Readers are warned not to take any action with regard to medical treatment without first consulting a physician. The TS Alliance does not promote or recommend any treatment, therapy, institution or health care plan.*

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