Genetic testing is one of several tools that doctors use to diagnose genetic conditions.Even when genetic testing is available, the tools listed above are used to narrow down the possibilities (known as a differential diagnosis) and choose the most appropriate genetic tests to pursue.The doctor may also use imaging studies including x-rays, computerized tomography (CT) scans, or magnetic resonance imaging (MRI) to see structures inside the body.The approaches to making a genetic diagnosis include: A physical examination: Certain physical characteristics, such as distinctive facial features, can suggest the diagnosis of a genetic disorder.Depending on the situation, specialized examinations such as nervous system (neurological) or eye (ophthalmologic) exams may be performed.Laboratory tests, including genetic testing: Molecular, chromosomal, and biochemical genetic or genomic testing are used to diagnose genetic disorders.A geneticist will do a thorough physical examination that may include measurements such as the distance around the head (head circumference), the distance between the eyes, and the length of the arms and legs.A personal medical history includes past health issues, hospitalizations and surgeries, allergies, medications, and the results of any medical or genetic testing that has already been done.Other laboratory tests that measure the levels of certain substances in blood and urine can also help suggest a diagnosis.In these cases, sequencing the entire genome may result in .locating the responsible genetic variant

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