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| Sommario/riassunto | "The time is fast approaching when virtually all the culprit genes and their mutations for 7,000 rare monogenic disorders1 will be known. Thus far causal single genes and their mutations have been determined for 5,6732 genetic disorders, enabling pre-implantation genetic testing or prenatal genetic diagnosis. These advances using chromosomal microarrays, whole exome sequencing and even whole genome sequencing together with fetal imaging, and non-invasive prenatal testing, expand the era in which all couples have the option of avoiding or preventing having children with irreversible, irremediable, crippling, or lethal monogenic disorders. Primary care physicians, and those in all medical specialties, will need to inform their patients of this key option. This imperative is already partly in current practice. Missing is the requirement of physicians to request and obtain the precise name of the genetic disorder in question or an existing DNA report on a family member, for prospective parents to benefit from available options"-- |