Condition: Diabetes mellitus

Inheritance:

Multifactorial; rare Mendelian forms exist.

Genetic etiology:

Type 1 diabetes: association with specific HLA haplotypes; polymorphism in insulin gene. Type 2 diabetes: multifactorial, with many genetic associations established. Mendelian disorders referred to as maturity-onset diabetes of the young, due to mutations in *GCK* gene encoding glucokinase, or other genes involved in control of insulin production or secretion.

Frequency:

Varies with different types and different populations.

Clinical features:

Diabetes mellitus is characterized by lack of production or responsiveness to insulin, resulting in chronic elevation of blood glucose. This leads to multiple complications, including obstruction of small blood vessels leading to blindness, peripheral neuropathy, renal disease, and cardiac dysfunction. Type 1 diabetes is due to autoimmune destruction of islet cells in pancreas; type 2 diabetes is due to insulin resistance, and is highly associated with obesity.

Management:

Type 1 diabetes treated by injection of insulin; type 2 diabetes treated with oral hypoglycemic drugs or insulin; both forms require careful attention to dietary management.

Genetic counseling:

Both forms multifactorial, with empirical data used for counseling.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.