The goals

How widespread is the problem?

How effective is treatment?
- Lifespan restored (completely corrective):
- Partial treatment:
**Why is treatment so ineffective?**

- mutant locus unknown
- irreversible pathology
- side effects

Best success:

...hence the drive to **find the genes**

**Possible points of intervention**

Mutant gene

\[\downarrow\]

Mutant mRNA

\[\downarrow\]

Mutant protein

\[\downarrow\]

biochemical dysfunction

\[\downarrow\]

Clinical phenotype

\[\downarrow\]

Family/Society
Detection

- Genetic counseling
  - Medical diagnosis – the need for accuracy
  - Pedigree analysis ➔ Risk estimate
  - Counseling/followup
**Prenatal or preimplantation testing**

- **Goals**

- **Methods**
  - Amniocentesis
  - Chorionic villus sampling
  - Preimplantation testing

- **Risks and ethical concerns**
Genetic screening

- Purpose

- Scope – who should be tested?

- Testing –
  - Deciding on a method
  - Pre-test and followup counseling

- Treatment options?
◊ Examples
  ◦ Screening for disease – PKU
  ◦ Screening for carrier status – sickle cell disease
Screening for carrier status – Tay-Sachs disease

Is it always appropriate to screen? – the CF example