

The goals

How widespread is the problem?

How effective is treatment?

- ◆ Lifespan restored (completely corrective):

- ◆ Partial treatment:

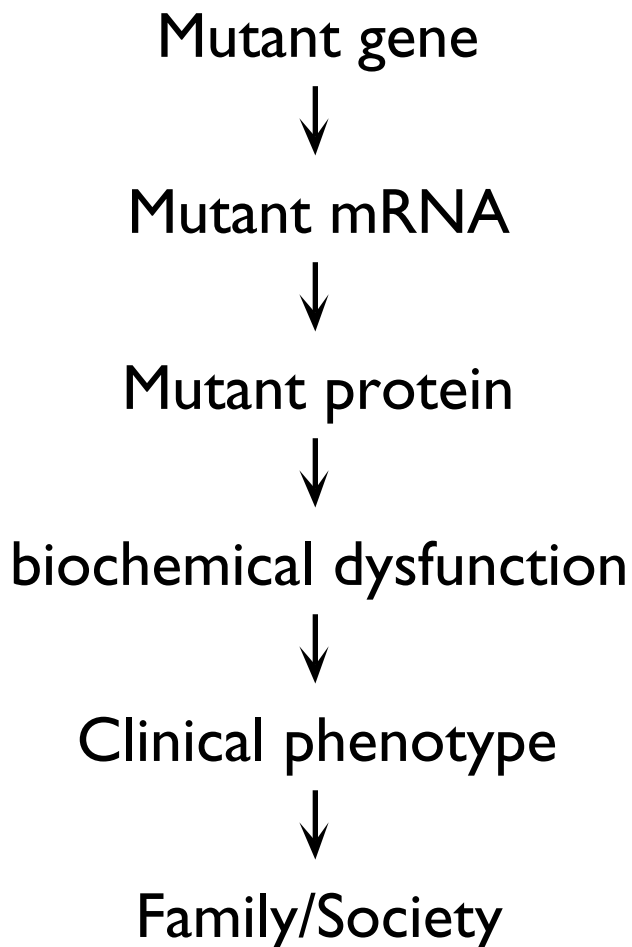
Why is treatment so ineffective?

- ◆ mutant locus unknown
- ◆ irreversible pathology
- ◆ side effects

Best success:

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

Possible points of intervention



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*