

Genetic Diseases

Krabbe Disease

- globoid cell leukodystrophy
- > Knud Haraldsen Krabbe
- impair growth and development of myelin sheath
- no cure yet

Dwarfism

- small stature
- autosomal dominant
- no treatment

Progeria

- accelerated human aging
- mutation in LMNA gene
↓
produce Lamin-A protein
- no cure

Fragile X Syndrome

- mutation in FMR1
- mental retardation
- no cure

Bioinformatics

- www.ncbi.nlm.nih.gov/

phenylketonuria

- phenylalanine hydroxylase deficiency

National Center for Biotechnology Information

- US National Library of Medicine

- resource for molecular biology information

Ensembl

- www.ensembl.org

- EMBL-EBI and Sanger Institute

- software system that produces and maintains automatic annotations on metazoan genomes.