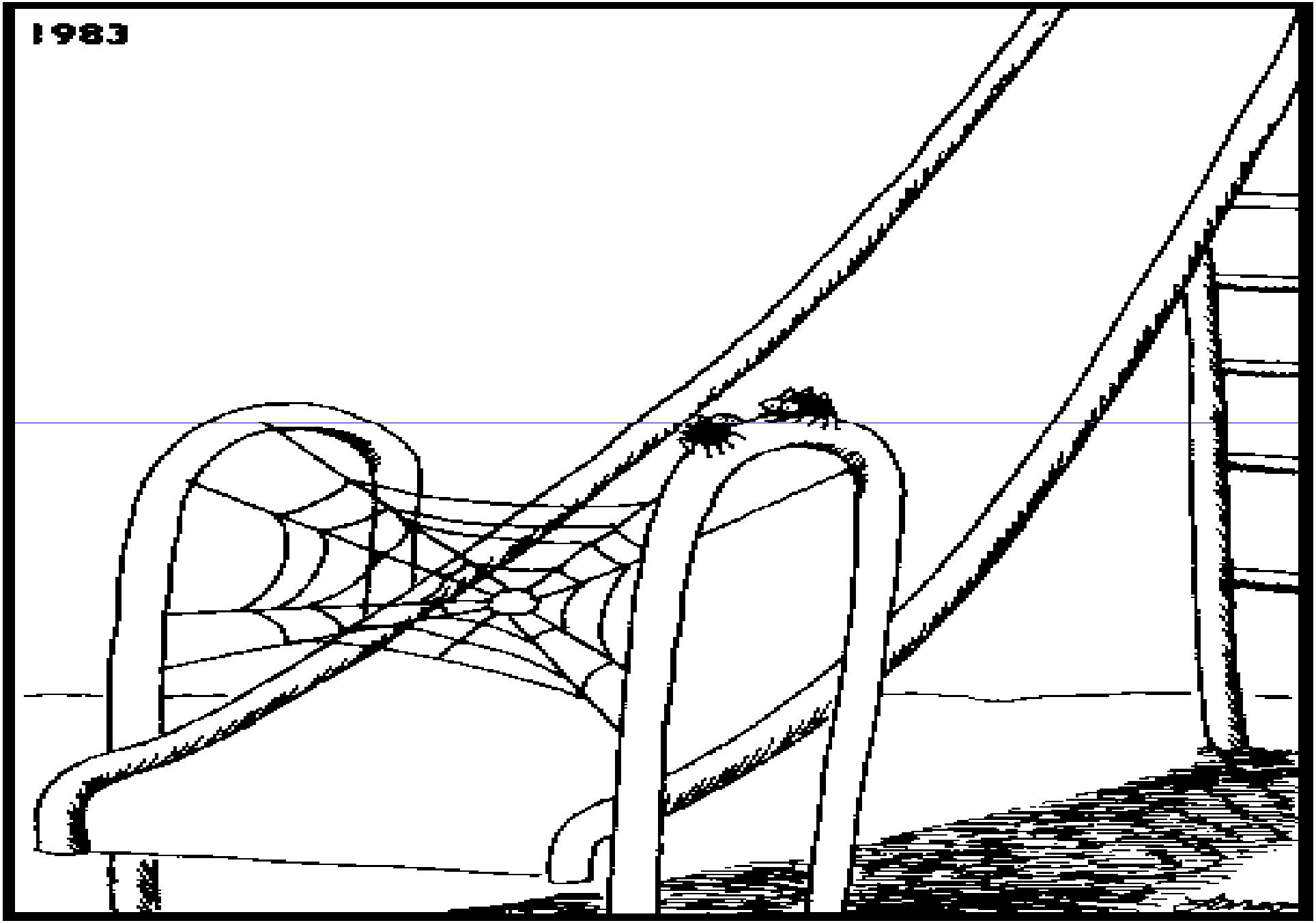
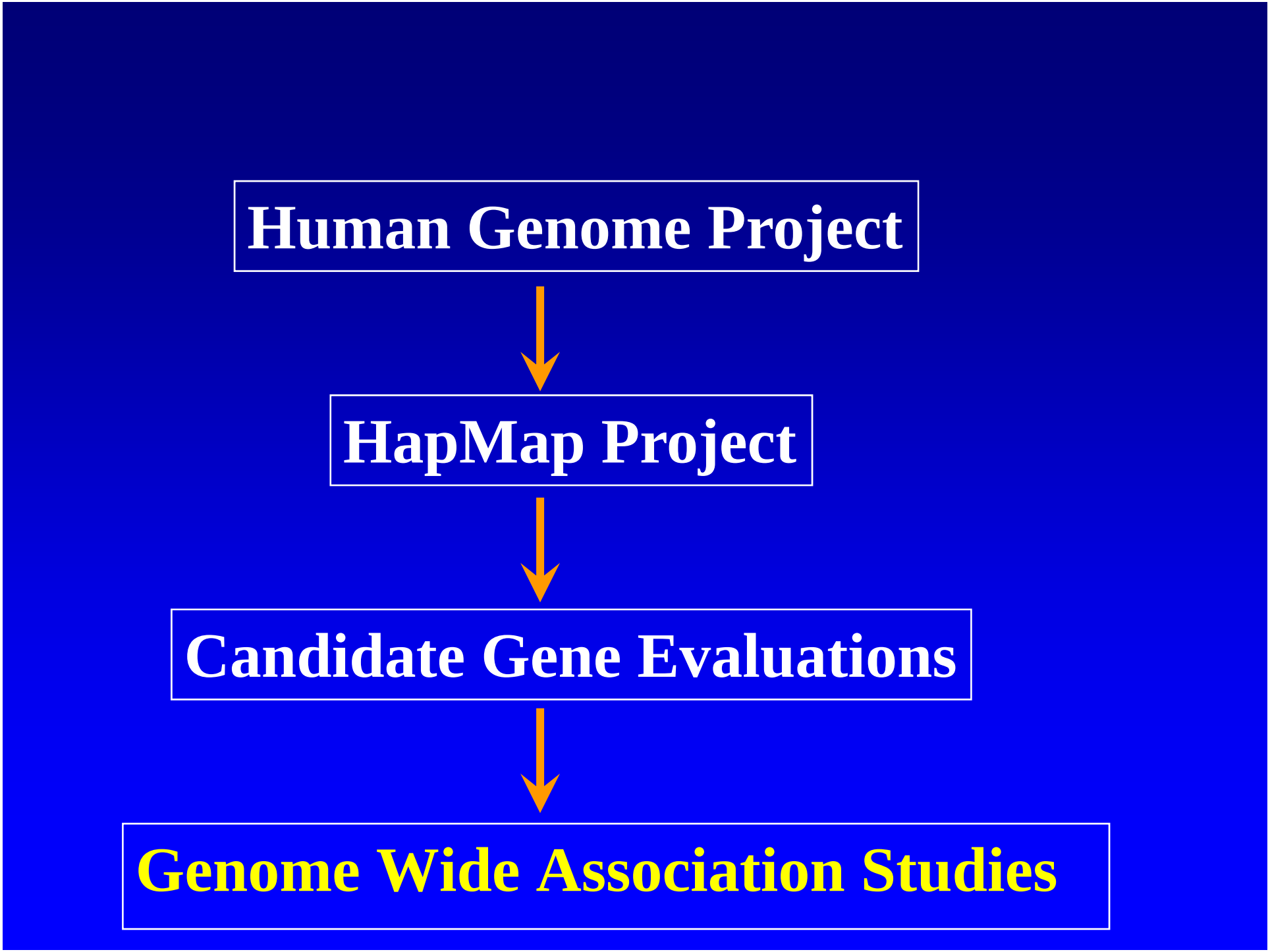


"If we pull this off, we'll eat like kings."

Large Cohort Studies as the "Population Laboratories" for Genetic Research

- Cohort Selection Allows Assessment of Larger Number of Risk Factors and Intermediate Factors
- Biologic repository is Collected **Prior** to Incident Event
- Ability to Assess **Change** in Risk Markers
- Reduced Bias in Outcome Assessment
  - (i.e., validation of outcomes, includes events with rapid mortality, can limit to **incident** events, etc)


Human Genome Project
HapMap Project
Candidate Gene Evaluations
Genome Wide Association Studies

Unprecedented Evolution in Cohort Studies During the Last Decade

Defining Genetic risk		**Data Analysis Issue**
Family history	→	Smaller Scale Local analyses
Targeted SNPs (N ~ tens)	→	Smaller Scale Local analyses
Larger # of SNPs (N ~ thousands)	→	Larger distributed analyses with required replication
GWA Studies (N ~ millionish)	→	Huge Data Issues: being made available to larger scientific community
Next Evolution?	→	**Next Evolution?**

Existing vs. New Cohorts?
Advantage of Existing Cohorts

- Cost Efficient
- Time Efficient (events available)
- Well Characterized Phenotypes
- Stored Biologic Specimens

Existing vs. New Cohorts
Challenges of Existing Cohorts

- **Data usually Disease Specific: often limited to common diseases (CA, CVD, DM, etc.)**
- **Consent from pre-GWA era**
- **No input on Data Collected/Outcomes**
- **Fewer Large Cohorts of Children**
- **Harmonizing phenotypes <u>across</u> cohorts**

Next Steps: Cohorts/Population Laboratories?

- **How can we enhance and standardize:**
  - Informed Consent/Participant Genetic Data Security
  - Harmonization of Exposures and Outcomes
  - Data Specimen Collection
    - frozen blood based specimens (DNA, Serum, etc) are inadequate for the "omics" revolution
    - what are needs of future population labs (e.g., tissue, fresh specimens, etc)?
- **How can we enhance use and publication of GWA data from existing Cohort Studies by the broader scientific community?**
- **When should new cohort studies be initiated?**
  - Should they span full age and outcome spectrum?
- **Should we embark on strategies to enhance efficiency?**
  - Initial use of case-control, medical record linkage studies, etc
  - Using existing well characterized cohorts to verify findings and better assess mediating factors?

What should be next for cohort studies?

- What are the priorities for the above issues?
- Who will lead these effort?
  - NHGRI?
  - Other?