RISK MODIFIERS IN THE DEVELOPMENT OF CONGENITAL HYDROCEPHALUS

Lance Lee, PhD is a faculty member in the Children’s Health Research Center at Sanford Research. Dr. Lee studies the genetic causes of primary ciliary dyskinesia, which has been linked to hydrocephalus. In his study, *Genetic Modifiers of Congenital Hydrocephalus*, Dr. Lee will study genes that modify the risk of developing hydrocephalus. By identifying these genetic modifiers, new drug targets may emerge to prevent or cure congenital hydrocephalus.

**GOAL**

Identify genes that modify the risk of developing hydrocephalus.

**THEORY**

People and mice are genetically heterogeneous. Some mice (and people) are at a higher risk of developing hydrocephalus. This difference in risk is linked to a person’s genetic makeup.

**EVIDENCE**

**MODEL: SAME MUTATION – DIFFERENT OUTCOMES**

This is dependent on the strain (genetic background).

**SAME GENETIC MUTATION**

- **Mild Ventriculomegaly**
- **Severe Ventriculomegaly**

How do we find the responsible genes?

1. **CONTROL**
   - Mate animals from different strains
2. Identify mice with varied levels of ventriculomegaly
3. Identify the genetic differences between these animals

**WHY IS THIS WORK INNOVATIVE?**

1. Identify new molecular pathways
   - New avenues for drug development
2. Identify genetic susceptibility
   - Potential for early risk assessment