



## **Holly S. Corbitt**

**Roche/ARCS Foundation Scholar – Portland Chapter**

**Recipient Institution:** Oregon Health & Science University

**Department:** Molecular & Medical Genetics

**Degree Pursuing:** PhD

**Adviser:** Dr. Cheryl Maslen

**Research Focus:** Holly is studying one of the major consequences of Down syndrome (DS), congenital heart defects. Atrioventricular septal defect (AVSD) occurs at a 2000 fold increase in DS, yet the genetic variants that cause this are still unknown. She is using sequencing data of DS kids with healthy hearts to those with AVSD to identify these genetic variants. She has identified 72 single nucleotide variants (SNVs) that are significantly associated with their cohort. Through whole exome sequencing, Holly has identified 48 genetic variants that could account for the increased risk of atrioventricular septal defects in children with Downs syndrome. An analysis of these genes showed a significant enrichment in the ciliome, which is a complex nano-machine. As a continuation of the study she will use this knowledge to elucidate the functional significance of the ciliome in early heart development and ultimately what causes it to go awry.

### **Publications/Presentations/Symposiums:**

- Program in Molecular & Cellular Biosciences retreat, Poster, 2014
- American Chemical Society Northwest Regional Meeting, Poster, 2011

### **Grants, Fellowships and Scholarships:**

- Roche/ARCS Foundation Scholar Award Program in the Life Sciences 2014
- NIH Ruth L Kirschstein T32 PMCB training grant recipient, 2014

**Undergraduate Education:** Portland State University, BS *summa cum laude* in molecular/microbiology and a minor in chemistry

### **Personal:**

- Mentor, Harrison Park Middle School
- City Club of Portland member
- Avid climber and swimmer