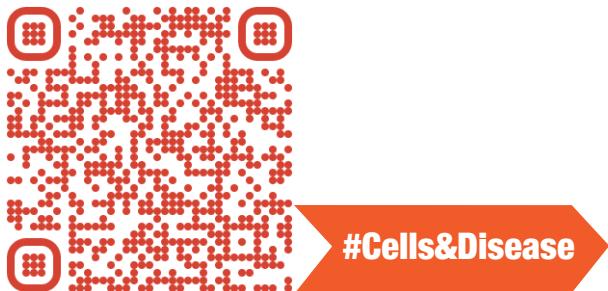


## CELL SIGNALING IN DEVELOPMENT AND DISEASE



Cell signaling is essential for development in multicellular organisms and when misregulated can lead to development defects and disease. This course examines several highly conserved cell signaling pathways in both model organisms and vertebrates and delves into a mechanistic understanding of pathway regulation using molecular, genetic, biochemical and cell biological approaches. There is a focus on dissecting key papers from the primary literature and learning about human defects and disease through class presentations.

### TOPICS

- Experimental approaches to study signal transduction
- Role of model organisms
- Notch signaling
- Wnt signaling
- Hippo signaling
- Hedgehog signaling
- Cilia in signaling
- Defects, diseases, and cancer in signaling pathway mutants

### INSTRUCTOR

Nancy Hawkins



Hiking enthusiast and  
Toronto Blue Jays fan

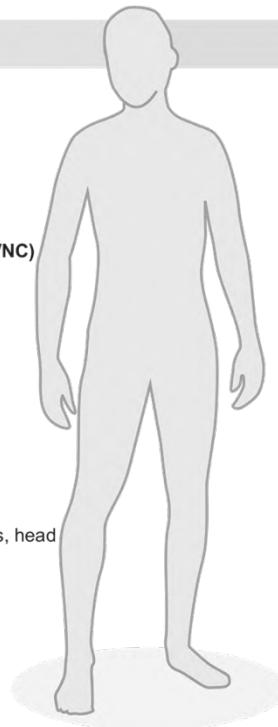


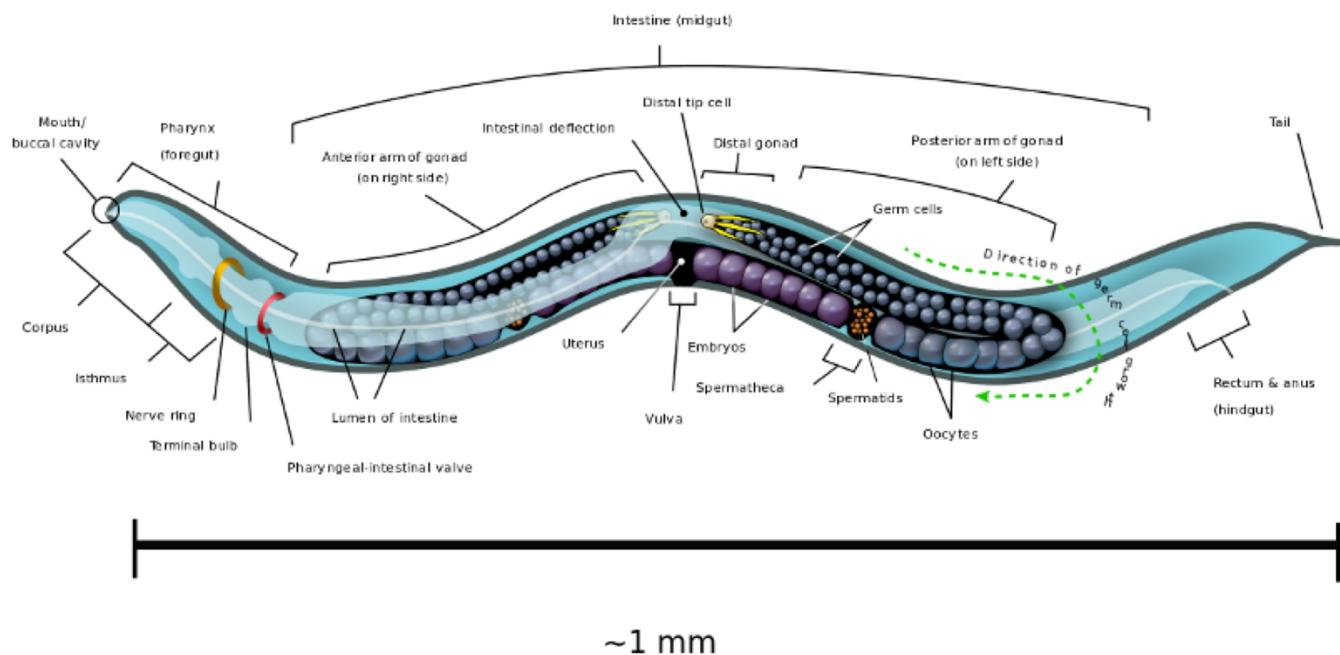
### Monogenic diseases with Notch pathway mutations

NOTCH1	<b>Bicuspid aortic valve disease</b> Heart
NOTCH1	<b>Hypoplastic Left Heart Syndrome</b> Heart
Mib	<b>Left Ventricle Cardiomyopathy (LVNC)</b> Heart
NOTCH1	<b>Thoracic aortic syndrome</b> Aorta
NOTCH1, CSL, DLL4	<b>Adams-Oliver syndrome</b> Skin, limbs
NOTCH2	<b>Hajdu-Cheney disease</b> Heart, nervous system, head
JAGGED1, NOTCH 2	<b>Alagille syndrome</b> Liver, heart, vasculature, kidney, lens, head
NOTCH3	<b>CADASIL</b> VSMC in brain
DLL3, LUNATIC FRINGE, HES7	<b>Spondylocostal dysostosis</b> Vertebral column

### Diseases with dysregulated Notch signaling

<b>Pulmonary Arterial Hypertension (PAH)</b> Lung, heart
<b>Chronic Obstructive Pulmonary Disease (COPD)</b> Lung
<b>Duchenne Muscular Disease (DMD)</b> Skeletal muscle
<b>Klippel-Feil syndrome</b> Vertebral column
<b>Pancreatitis</b> Pancreas
<b>Fibrosis</b> Lung, liver, heart





~1 mm

