“Synergy between genetic studies in patients and experimental animals: Hypopituitarism and disorders of sexual development”

Camper Lab

Corepressors TLE1 and TLE3 interact with HESX1 and PROP1


Department of Human Genetics, University of Michigan, Ann Arbor, Michigan 48109, USA.
Identification of stem cell markers in the pituitary gland of transgenic mice with hypopituitarism (Financial Support from FAPESP)

A novel heterozygous OTX2 deleterious variant (c.689 C>T; p.H230L) in a patient with hypopituitarism and ectopic posterior pituitary without eye malformation

*Functional Study*

Exome analysis/ Sanger confirmation

Identification of novel variants via exome sequencing in patients with isolated or combined hormone deficiencies (sporadic, familial, consanguineous)

Setting up a course: Animal Models as Tools for investigating Endocrine Disease

One week teaching about Transgenic Animal Techniques
Second round focused in Zebrafish: Animal Models as Tools for investigating Endocrine Disease

One week teaching about zebrafish Techniques

Disorders of Sexual Development Collaboration

7th Annual Pituitary – Hypothalamus Meeting (Brains and Hormones)
October 11-13, 2013
Kellogg Biological Station, Gull Lake, Michigan, USA

Sorahia Domenici

Catherine Keegan

Topics for Discussion in the roundtable

1) Briefly note how collaboration began and what initial research questions addressed together were; IESP

2) Provide brief overview of one or two key challenges to developing collaboration and how addressed and describe evolution of research questions addressed together; Work as a team!

3) Highlight current priorities-, research topics being addressed together and projected next steps in the short and longer term; Exome analysis to find out candidate genes, test in vitro and animal models our hypothesis and publish

4) Conclude with one or two lessons from work together that may be useful for participants working to develop collaborations between UM-Brazil regardless of the topic area.-
   - Work as a team!
   - Knowing Brazilian difficulties could help us getting supplies from our international collaborator!

Thank you