The Challenges and Opportunities of Collaboration

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Disorders @ CSC

132 known variants, 110 disorders

5 benign disorders

3 de novo dominant

2 still under review

122 known variants, 100 disorders

Recessive: 107 variants, 85 disorders

Dominant: 10 ; X-linked: 5

Identify about 10 new variants/year
Amish
78 variants
GA-1, galactosemia
GM3 synthase def.

Mennonite
47 variants
MSUD, SMA, MCAD

Both
7 variants
5%
PA, F5

Same disorders, different variants: SCID, PKU
Why Collaborate?
CSC Weaknesses

- Functional protein studies
- Model organism work
- Staff hearing booth, cardiology equipment, ophthalmology
- Perform exome and genome sequencing
- Many biochemical assays
- Investigate each disorder in detail
CSC Strengths

• Clinical
  • Patients with disorders, frequently serious and life threatening
  • Provide primary care, need treatments/protocols, develop and test
  • Clinical trials
  • Always something new to study

• Lab
  • Gene discovery (microarrays, gene sequencing, exome data analysis)
  • Biochemical studies (GC-MS, ELISA, HPLC)
  • Diagnostic services
  • Hearing booth, cardiology equipment, ophthalmology equipment
  • Understanding of the population, institutional knowledge (educate others)
Why Collaborate?

- We can’t do it all (skills, equipment, staff)
- We can’t know it all
- Need input from other individuals and organizations to help strengthen a project
- New opportunities
- Advance research (patient care) in a way we can’t on our own
- Fill weaknesses in our organization with strengths in another (vice versa)
Collaborators

• #1: Research is patient focused
• Select collaborators carefully
  • Good working relationship
• Communicate
• What are the manpower requirements?
• Know your limits, you can’t work with everyone!
Projects @ CSC

- CNTNAP2 - better phenotyping - oxytocin trial
- GM3 synthase deficiency - G500 trial - DDC, CCRC, Hess lab, Fonterra, Nemours
- Heredity thromboembolism - gene discovery, functional data - Jinks Lab, Amish Research Center
- Formula design (PKU, MSUD, PA) - Applied Nutrition
- Ion Torrent PGM - CSC panel validation
- Hearing loss - Nemours
- Various patient specific projects
GeneX, bipolar disorder
Bucan Lab – U. Penn
Hoshi Lab – U. Penn
Markx Lab – Columbia U.
First Lab – Columbia U.
Paul Lab – Weill Cornell
Ann McDonald, MD

HARS, Usher syndrome 3B
Francklyn Lab - UVM Med

HERC2, Autism spectrum disorder
Center for Special Needs Children

GeneZ, CODAS syndrome
Superi-Furga Lab – U. Lausanne
Innes Lab – U. Calgary
Deardorff Lab – U. Penn/CHOP
Lee Lab – Case Western Reserve U.
Suzuki Lab – UMDNJ

FLVCR1, posterior column ataxia and retinitis pigmentosa
Higgins Lab – Athena Diagnostics

SLITRK6, congenital sensorineural hearing loss
Crino Lab – Temple Med. School
A.I. duPont Children’s Hospital

GeneY, Yoder dystonia
Hildebrandt Lab – Boston Children’s

Adapted from a presentation by Rob Jinks, PhD, Franklin & Marshall College
Franklin & Marshall College

- Unique student opportunities and education
- Funding for CSC and F&M (institutions, individual students)
- Real world problems
- Real world context
- Unique research and publications (student authors)
- Interaction with patients
- Good way to publicize work
- Unique educational opportunities, viewpoint
- Research into CSC disorders, benefit to patients

HHMI - Brigatti

CSC F&M

Functional Protein Studies - Jinks
Public Health Initiatives - Miller, Rice, Yost
GM3 synthase deficiency - Hess, Wesalo
Thank You!