OREGON HEALTH & SCIENCE UNIVERSITY

Rare Disorders Research Consortium (RDRC)

OHSU Research Consortium

OHSU RDRC Overview

The Rare Disorders Research Consortium (RDRC) was established to act as a central knowledge resource with the goal of partnering researchers with stakeholders in the rare disease community including other researchers, patient advocates, clinical & regulatory organizations, funding organizations and drug companies.

OHSU rare disease research spans the entire translational pipeline, including disease gene discovery, mechanisms, animal, cell, and human model testing, research resources, innovative diagnostics and profiling, preclinical studies, and clinical trials. OHSU faculty & staff perform basic research, diagnose and treat around ninety rare diseases.

We are actively seeking partnerships in academia and industry to advance our goal to discover and apply innovative diagnostics and effective therapeutics for our portfolio of rare diseases, including rare cancers.

Rare Disorders Tests Developed with OHSU Collaborators

- Fanconi anemia (FANCA,C,G,E,F)
- NBIA (PANK2, PLA2G6, FA2H, MMIN, ATP13A2)
- Carnitine palmitoyltransferase 1A deficiency (CPT1A genotyping)
- Achromatopsia (CNGA3, CNGB3)
- Rett (MeCP2, CDKL5)
- Cystic fibrosis (CFTR)
- MEN2A, MEN2B, FMTC (RET proto-oncogene)
- Noonan (PTPN11)
- DFNB1 nonsyndromic hearing loss and deafness (GJB2 - Connexin 26)

http://www.ohsu.edu/rdrc

Resources

- Animal models including the Oregon National Primate Research Center for developing therapeutics and safety studies
- Access to patient databases, clinical trials and outcomes research, and the Newborn Screening Translational Research Network
- A unique collection of research and clinical materials, including annotated archival materials and primary cells
- Strong infrastructure for design and execution of clinical trials, including the Oregon Clinical & Translational Research Institute
- Experience and proven track record for successful completion of all phases of clinical trials
- **Diagnostics** (CLIA and CAP certified) translates new disease genes into clinical tests and conducts technology evaluation, including platform validation for clinical studies. **Technologies include:** Sequencing, deletion/ duplication analysis, genotyping, sterol analysis.

Research Strengths

- Nationally distinctive capabilities in neurodegeneration with brain iron accumulation (NBIA), Fanconi Anemia, sterol disorders, ophthalmologic disorders, chronic myelogenous leukemia (CML), connective tissue disorders, Fatty Acid Oxidiation disorders, Urea Cycle disorders, and Huntington's disease
- Clinical trials designed and conducted specifically for rare disorders
- Joint pre-clinical research programs in target areas

National Rare Disease Clinical Research Consortiums: Primary Immune Deficiency Treatment Consortium, Brittle Bone Disorders, Sterol and Isoprenoid Disorders, Urea Cycle (affiliate center)

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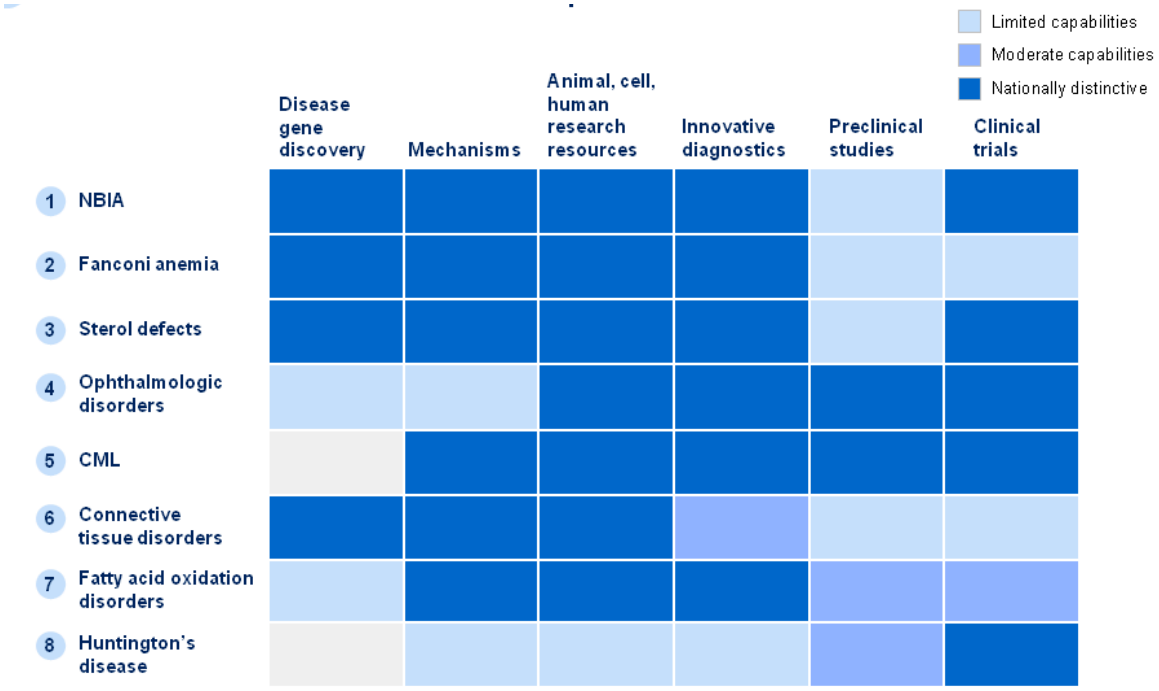

Selected Major Expertise Areas

Cancer susceptibility disorders: Fanconi anemia, mismatch repair cancers, xeroderma pigmentosum (XP), PTEN hamartoma tumor syndrome (PHTS), familial melanoma syndrome, rhabdomyosarcomas

Neurodegenerative disorders: Neurodegeneration with brain iron accumulation (NBIA), Huntington's disease, long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency

Ocular disorders: Retinitis pigmentosa, Stargardt disease, uveitis, Leber's congenital amaurosis, retinoschisis, achromatopsia, macular degeneration

Sterol metabolic defects: Smith-Lemli-Opitz Syndrome (SLOS), Sitosterolemia, Mevalonate Kinase Deficiency (MKD/HIDS), Sjögren-Larsson Syndrome (SLS), Methylsterol Oxidase Deficiency (SC4MOL), Cerebrotendinous Xanthomatosis (CTX)

Bone & connective tissue disorders: Marfan syndrome, osteogenesis imperfect (OI), Weill-Marchesani syndrome

Inborn Errors: Tyrosinemia, CPT1a deficiency, phenylketonuria (PKU)

Protein misfolding defects: Cystic Fibrosis, HERG, Hyperinsulinemia of infancy (SUR), many others

Renal Transport: pseudohypoaldosteronism type 2, familial hyperkalemic hypertension, Gitelman syndrome, Bartter

RDRC leadership and contact information

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