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NIH Announces Expansion of Rare Diseases Clinical Research Network

Nineteen New and Returning Consortia to be Awarded \$117 Million

The National Institutes of Health announced today a second phase of the Rare Diseases Clinical Research Network (RDCRN) including funds for 19 research consortia. The Rare Diseases Clinical Research Consortia and a Data Management Coordinating Center (DMCC) will be awarded a total of just over \$117 million over the next five years. The research conducted with the new funding will explore the natural history, epidemiology, diagnosis, and treatment of more than 95 rare diseases.

"The progress made by researchers through the network over the past six years is important and impressive," said NIH Director Francis S. Collins, M.D., Ph.D. "We have shown that this approach can be a catalyst for progress in meeting the challenge of rare diseases, and we are eager to launch this next phase of the program."

A rare disease is defined as a disease or condition affecting fewer than 200,000 persons in the United States. Approximately 6,500 such disorders have been identified, affecting an estimated 25 million Americans.

Initially created in 2003, the RDCRN is unique in its approach to addressing rare diseases as a group. Previously, the NIH's institutes and centers funded research on individual rare diseases in their respective disease-type or organ domains. The RDCRN is the first program that aims to create a specialized infrastructure to support rare diseases research.

Since its creation, the RDCRN has enrolled over 5,000 patients in 37 clinical studies in rare diseases. Patient recruitment for clinical studies is a fundamental challenge in rare diseases research because there are typically so few affected patients in any one area. The RDCRN was designed to address this problem by fostering collaboration among scientists and shared access to geographically distributed research resources. Network consortia have also established training programs for clinical investigators who are interested in rare diseases research.

"Collaboration is a critical element of rare diseases research and the partnerships represented in this program have tremendous potential to make great strides in understanding these diseases," said Stephen C. Groft, Pharm.D., director of NIH's Office of Rare Diseases Research (ORDR). "The network emphasizes collaboration not just among investigators from multiple research sites but between investigators and patient advocates as well."

The direct involvement of patient advocacy groups in network operations, activities, and strategy is a major feature of the RDCRN. Each consortium in the network includes relevant patient advocacy groups in the consortium membership and activities. These patient advocacy group representatives serve as research partners within their own consortia. Collectively, the Coalition of Patient Advocacy Groups

(CPAG) represents the perspective and interests of all patient advocacy organizations associated with the RDCRN. The CPAG participants meet frequently throughout the year via teleconference and face-to-face meetings. They participate in network-level discussions and meetings. The CPAG chairperson is a voting member of the RDCRN Steering Committee.

Funds and scientific oversight for the RDCRN will be provided by ORDR and seven NIH Institutes, which will also contribute considerable administrative support to the network: the National Institute of Neurological Disorders and Stroke (NINDS), the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), the National Institute of Allergy and Infectious Diseases (NIAID), the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), the National Institute of Dental and Craniofacial Research (NIDCR), the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK), and the National Heart, Lung and Blood Institute (NHLBI). Several consortia will also receive financial support from their associated patient advocacy groups.

In the RDCRN's first phase, the network's Data and Technology Coordinating Center (DTCC) developed a management system for the collection, storage, and analysis of RDCRC data, and additional systems to address needs of individual studies, such as a laboratory data collection system, a specimen tracking system, and a pharmacy management system (to support blinded distribution of study agents and placebos). The DTCC also created RDCRN's central public Web site, developed as a portal for the rare diseases community, including patients and their families and health care professionals, to provide information on rare disease research, consortium activities, RDCRN-approved protocols, disease information, and practice guidelines. Located at <http://rarediseasesnetwork.epi.usf.edu/>, the Web site had over 3.4 million visits in 2008. The RDCRN DTCC also developed a unique voluntary patient registry that provides ongoing contact with approximately 5,000 individuals from over 60 countries representing 42 diseases, alerting them when new studies are opened in the network or when ongoing studies expand to new sites.

In this second phase of the RDCRN, the University of South Florida will continue these data management efforts, under a new name and with a slightly different charge, as the Data Management Coordinating Center (DMCC). The DMCC will develop uniform investigative clinical research protocols for data collection in collaboration with the RDCRN Steering Committee, monitor protocol adherence, data collection and data submission, and work with the each consortium's Data and Safety Monitoring Boards to establish protocols for adverse events notification and reporting.

"This innovative program provides unique insights into the development of rare diseases as well as therapeutic opportunities," according to Story C. Landis, Ph.D. director of the NINDS. "The NINDS is proud to administer the RDCRN data management coordinating center on behalf of the NIH."

The 19 consortia and DMCC included in this second phase of the RDCRN are listed below.

| Consortium Title, Institution, and Principal Investigator | NIH Collaborators | Diseases to be studied |
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| Angelman, Rett, and Prader-Willi Syndromes Consortium - University of Alabama at Birmingham - Alan | ORDR, NICHD | Angelman syndrome, Rett syndrome, Prader-Willi syndrome |

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| K. Percy, M.D. | | |
| Autonomic Rare Diseases Clinical Research Consortium - Vanderbilt University - David Robertson, M.D. | ORDR, NINDS | Multiple system atrophy (MSA), baroreflex failure, autoimmune autonomic neuropathy, pure autonomic failure (PAF), hypovolemic postural tachycardia syndrome (hPOTS), dopamine beta hydroxylase deficiency (DBHD) |
| Brain Vascular Malformation Consortium - University of California, San Francisco - William L. Young, M.D. | ORDR, NINDS | Vascular malformations: cerebral cavernous malformation progression, Sturge-Weber syndrome, hereditary hemorrhagic telangiectasia |
| Clinical Investigation of Neurologic Channelopathies (CINCH) - University of Rochester - Robert C. Griggs, M.D. | ORDR, NINDS | Nervous system channelopathies: Andersen-Tawil syndrome, episodic ataxias, non-dystrophic myotonic disorders |
| Dystonia Coalition - Emory University - Hyder A. Jinnah, M.D. | ORDR, NINDS | Focal dystonias, cervical dystonia, blepharospasm, spasmodic dysphonia, craniofacial dystonia, limb dystonia |
| Genetic Disorders of Mucociliary Clearance - University of North Carolina at Chapel Hill - Michael R. Knowles, M.D. | ORDR, NHLBI | Primary ciliary dyskinesia (PCD), cystic fibrosis (CF), pseudohypoaldosteronism (PHA) |
| Hereditary Causes of Nephrolithiasis and Kidney Failure | ORDR, NIDDK | Rare hereditary stone diseases: primary hyperoxaluria, cystinuria, dihydroxyadeninuria, Dent's disease |

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| <p>- Mayo Clinic College of Medicine, Rochester - Dawn S. Milliner, M.D.</p> | | |
| <p>Immune Mediated Disorders After Allogeneic Hematopoietic Stem Cell Transfer - Fred Hutchinson Cancer Research Center - Stephanie J. Lee, M.D., M.P.H.</p> | <p>ORDR, NIAID</p> | <p>Cutaneous sclerosis, bronchiolitis obliterans, late acute graft versus host disease (GVHD)</p> |
| <p>Inherited Neuropathies Consortium - Wayne State University - Michael E. Shy, M.D.</p> | <p>ORDR, NINDS</p> | <p>Inherited peripheral neuropathies: Charcot-Marie-Tooth diseases (CMT) including 1) CMT1, the dominantly inherited demyelinating neuropathies, 2) CMT2, the dominantly inherited axonal neuropathies, 3) CMT4, the recessively inherited neuropathies</p> |
| <p>Lysosomal Disease Network -University of Minnesota Twin Cities - Chester B. Whitley, M.D.</p> | <p>ORDR, NINDS, NIDDK</p> | <p>Lysosomal diseases: mucopolysaccharidosis (MPS), MPS bone disease, Pompe disease, Niemann-Pick disease type C, glycoproteinoses, Wolman disease, late infantile ceroid lipofuscinosis, (LINCL), mucopolipidosis type IV, hexosaminidase deficiency, Fabry disease nephropathy, Batten-Turner muscular dystrophy</p> |
| <p>Molecular and Epidemiologic Characterization of Salivary Gland Carcinomas - University of Texas M.D. Anderson Cancer Center - Adel K. El-Naggar, M.D., Ph.D.</p> | <p>ORDR, NIDCR</p> | <p>Salivary gland carcinomas: mucoepidermoid carcinoma (MEC), adenoid cystic carcinoma (ACC), adenocarcinoma (ACC)</p> |
| <p>Nephrotic Syndrome Rare Disease Clinical Research Network - University of</p> | <p>ORDR, NIDDK</p> | <p>Focal and segmental glomerulosclerosis (FSGS), minimal change disease (MCD) and membranous nephropathy (MN)</p> |

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| <p>Michigan, Ann Arbor - Matthias Kretzler, M.D.</p> | | |
| <p>North American Mitochondrial Diseases Consortium - Columbia University Medical Center - Salvatore DiMauro, M.D.</p> | <p>ORDR, NINDS</p> | <p>Mitochondrial encephalopathy lactic acidosis with stroke-like episodes (MELAS); mitochondrial neurogastrointestinal encephalomyopathy (MNGIE); Leber's hereditary optic neuropathy (LHON), LHON and dystonia, Leigh syndrome; encephalomyopathy; ALS-like syndrome of encephalomyopathy; neuropathy, ataxia and retinitis pigmentosa syndrome (NARP); maternally inherited Leigh syndrome (MILS); familial bilateral striatal necrosis (FBSN); leukodystrophy; CoQ deficiency; encephalopathy; cardioencephalomyopathy; leukodystrophy/tubulopathy; fatal infantile encephalomyopathy</p> |
| <p>Porphyria Rare Disease Clinical Research Consortium - Mount Sinai School of Medicine of New York University - Robert J. Desnick, Ph.D., M.D.</p> | <p>ORDR, NIDDK</p> | <p>Porphyrias: Acute Intermittent Porphyria (AIP), variegate porphyria (VP), hereditary coproporphyria (HCP), aminolevulinatase deficiency porphyria (ADP), porphyria cutanea tarda (PCT), erythropoietic protoporphyria (EPP), congenital porphyria (CP)</p> |
| <p>Primary Immune Deficiency Treatment Consortium - University of California, San Francisco - Morton Cowan, M.D.</p> | <p>ORDR, NIAID</p> | <p>Primary immune deficiencies: severe combined immunodeficiency (SCID), Wiskott-Aldrich syndrome (WAS) and chronic granulomatous disease (CGD)</p> |
| <p>Spinocerebellar Ataxia - Clinical Research Consortium - University of Florida, Gainesville - Tetsuo Ashizawa, M.D.</p> | <p>ORDR, NINDS</p> | <p>Spinocerebellar ataxia: SCA 1, 2, 3, and 6</p> |
| <p>Sterol and Isoprenoid Diseases Consortium -</p> | <p>ORDR, NICHD</p> | <p>Niemann-Pick disease type C, Smith-Lemli-Opitz syndrome, Sjögren-Larsson syndrome, mevalonate kinase deficiency, hyper-IgD syndrome, cerebrotendinous xanthomatosis (CTX),</p> |

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| Oregon Health and Science University - Robert David Steiner, M.D. | | sitosterolemia |
| Urea Cycle Disorders Consortium - Children's National Medical Center Research Institute - Mark L. Batshaw, M.D. | ORDR, NICHD | Urea cycle disorders: N-acetylglutamate synthetase (NAGS) deficiency, carbamoyl phosphate synthase 1 (CPS) deficiency, ornithine transcarbamylase (OTC) deficiency, argininosuccinate synthetase deficiency (classic citrullinemia), citrin deficiency (citrullinemia type 2), argininosuccinate lyase deficiency (argininosuccinic aciduria), arginase deficiency (hyperargininemia), ornithine translocase deficiency syndrome (HHH) |
| Vasculitis Clinical Research Consortium - Boston University Medical Campus - Peter A. Merkel, M.D., Ph.D. | ORDR, NIAMS | Vasculitides: Wegener's granulomatosis (WG), microscopic polyangitis (MPA), Churg-Strauss syndrome (CSS), polyarteritis nodosa (PAN), Takayasu's arteritis (TAK), giant cell (temporal) arteritis (GCA) |
| Data Management and Coordinating Center (DMCC) - University of South Florida - Jeffrey C. Krischer, M.D. | ORDR, NINDS | |

The NIH Office of Rare Diseases Research (ORDR) stimulates and coordinates research on rare diseases and supports research to respond to the needs of patients, healthcare providers and the research communities involved in efforts to improve the lives of patients and families facing rare diseases. For more information about ORDR and its programs, visit <http://rarediseases.info.nih.gov/>.

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