Analysis of NIDCR's Rare Disease Portfolio

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What are “Rare” diseases?
• Diseases which are characterized by a low prevalence (<200,000 people) in the population. They frequently are associated with problems in diagnosis and treatment.
• But, having a rare disease is not so rare. Of the roughly 6,800 rare diseases that are known, about 10% of the US population has some form of rare disease.
• There are more than 5,000 rare disorders that, taken together, affect approximately 20 million Americans.
• One in every 12 individuals in this country has received a diagnosis of a rare disease (from National Organisation for Rare Disorders).

Funded research is heavy in basic sciences, with few pre-clinical studies. More than half the projects are related to head and neck cancer.

Abstract
In order to understand the landscape of rare diseases affecting the dental, oral, and craniofacial skeleton, an analysis was conducted of NIDCR’s research grants in FY14. Using manually coded descriptors of NIDCR’s funded research based on reading the applications abstracts, a broad categorization was conducted. The aim was to gain an understanding of the current portfolio with regard to the disease research, and to explore needs, gaps, and opportunities in this area. Using analytical tools, additional examination of the research outputs, key players, and the impacts of these outputs are presented. By identifying the hurdles, successes, and state of progress towards clinical readiness in rare diseases, the NIH will be better poised to leverage resources for funding rare disease research in the area of dental, oral, and craniofacial science. The ultimate goal is to look at scientific areas where rare disease research has helped advance knowledge in related, but more common, diseases and pathways.

More than half the projects are related to head and neck cancer.

Articulating landscape and opportunities

Disease categorization by ORDR framework

The Hypophosphatasia Story
Asfotase alfa as an enzyme replacement therapy

Summary
• Funded research is heavy in basic sciences, with few pre-clinical studies.
• There are no clinical trials in NIDCR portfolio.
• More than half the projects are related to head and neck cancer.

Future Analyses
• Are there other rare disease areas we should be funding?
• What are the hurdles in advancing rare disease research?
• Has rare disease research helped advance discoveries or treatment of more prevalent diseases?