Rare Disease as an Emerging and Distinct Field of Medicine

With the completion of the human genome sequence and the advent of affordable molecular sequencing, the field of genetic rare diseases has exploded. With over 8000 conditions described (and counting), the classic expectation of a physician's encyclopedic knowledge of their field is unrealistic. The pace of therapeutic development for rare disease is also explosive with 52% of new drugs approved by the FDA for rare diseases. This lecture will describe this oncoming wave and discuss new models that can be used to care for patients.