Critical mass in rare diseases -- an innovative Internet approach

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The internet is emerging as a valuable tool for scientists to gather data for critical research into rare diseases. Disability researcher, Dr Helen Leonard, from Australia's Institute for Child Health Research, said its InterRett online database had greatly expanded research into the neurological disorder Rett syndrome, which affects 1 in 8500 girls.

"In a rare disease like Rett syndrome, with considerable clinical and genetic heterogeneity, there is an urgent need and value for combining data from different sources through initiatives such as the international database InterRett, the IRSF Rett Phenotype Database," Dr Leonard said.

"By having a web-based database, families and clinicians from around the world can contribute their information and help us to achieve a critical mass of data that begins to make statistically relevant analysis possible."

The benefits and future potential of InterRett will be showcased at the World Rett Syndrome Congress to be held in Paris, France from October 10.

As of September 2008, InterRett had ascertained 1691 cases of Rett syndrome. These cases came from 39 different countries, with the largest proportion coming from Australia (21%), Spain (21%), USA (18%), France (13%), Israel (5%), China (5%) and the UK (5%).

Funded by the International Rett Syndrome Foundation (IRSF), InterRett examines the clinical features and genetic characteristics of Rett syndrome.

Dr Leonard said that for rare disorders such as Rett syndrome, the Internet provides access to a worldwide population, providing higher statistical power than individual centres or even country based research studies.

"Just as importantly, the internet also provides an ideal medium to disseminate high quality information about a rare disorder to the medical and general community," she said.

Dr Leonard said a range of issues surrounding internet databases will be discussed at the Congress including the need for and development of standard data collection tools; research questions which can be answered using pooled data; the role of parent support organisations in promoting family participation in InterRett; technical issues relating to the pooling, de-identification of data whilst still avoiding duplication of cases; and funding strategies to better support the data custodians and their analytic capacity and to facilitate the "pooling" process.

Dr Leonard said she was hopeful that similar internet databases would be developed for other rare diseases.

"Rare diseases collectively affect up to 10 percent of the population and it's so important that we can gather accurate data to inform clinical practice, government policy, and health service planning," she said.

The breadth and scope of the 2008 Congress will be a first for Rett syndrome. It will extend from a detailed exploration of the neurobiology of Rett syndrome, particularly incorporating the work being done on mouse models to basic clinical and epidemiological research relating

clinical presentation to genetic determinants.

There will also be a focus on potential new therapeutic options and the best way to undertake clinical trials where these and other therapies can be assessed.

Source: Research Australia