

## **Orphan Mechanisms of Primary Ovarian Insufficiency: Passion for Participatory Research**

October 2 - 3, 2008  
Bolger Center, Potomac, Maryland

### *Summary*

In October 2008 Rachel's Well, Inc., the NIH Intramural Research Program, the NIH Office of Research on Women's Health, the DHHS Office of Women's Health, and the NIH Office of Rare Diseases collaborated to organize a scientific conference titled, "Orphan Mechanisms of Primary Ovarian Insufficiency: Passion for Participatory Research." The goal was to get all of the stakeholders in one room, search out common goals, and to initiate a process that would facilitate working together to raise awareness, remove barriers to care, and stimulate research in the interest of all women who experience primary ovarian insufficiency (POI). There were several "orphan conditions" and communities represented. The following communities have primary ovarian insufficiency as a part of their problem list: families affected by fragile X syndrome, galactosemia, Turner syndrome, disorders of sexual development due to sex chromosome abnormalities, autoimmunity, lupus, cancer chemotherapy, and the many cases in which no cause can be found.

A team of representatives, including members of Rachel's Well, Inc., is developing a written report summarizing the meeting. The main conclusion from the meeting was that there is a need for a POI research consortium. A report to be published in a peer reviewed journal will articulate a strategy for making this a reality. Following the meeting the community has formed working groups to advance various components of the consortium. One of these, the "POI Phenotype and Clinical Trials Database Working Group," took on as its major agenda to develop a network of POI clinics that will provide integrated care across the nation for these women as part of a patient registry. These patients need help and guidance with their emotional health, their endocrine health, their genetic health, and their reproductive health. This requires integrated care, which the patients who participated in the conference told us is difficult for them to find in the current configuration of our health care system. In a parallel effort this working group is developing a standardized database for these patients, so that economy of scale can become a research ally here.

The "POI Family Planning Working Group," also formed as a result of this meeting, is developing a research agenda to deal with the emotional health of these women. This team includes representation from positive psychology, spiritual ministry, occupational therapy, and reproductive psychiatry. The "POI Genetics and Epidemiology Working Group" that formed as a result of this meeting is initiating collaboration between NIH and the Agency for Healthcare Research and Quality. Plans are in the making to hold a follow up meeting to examine the feasibility of forming a patient registry and research effort that employs the basic principles of community based participatory research.

So, as a result of this conference a POI community has formed. The community is working together to raise awareness, remove barriers to care, and stimulate research in POI and the rare disorders that relate to this condition.