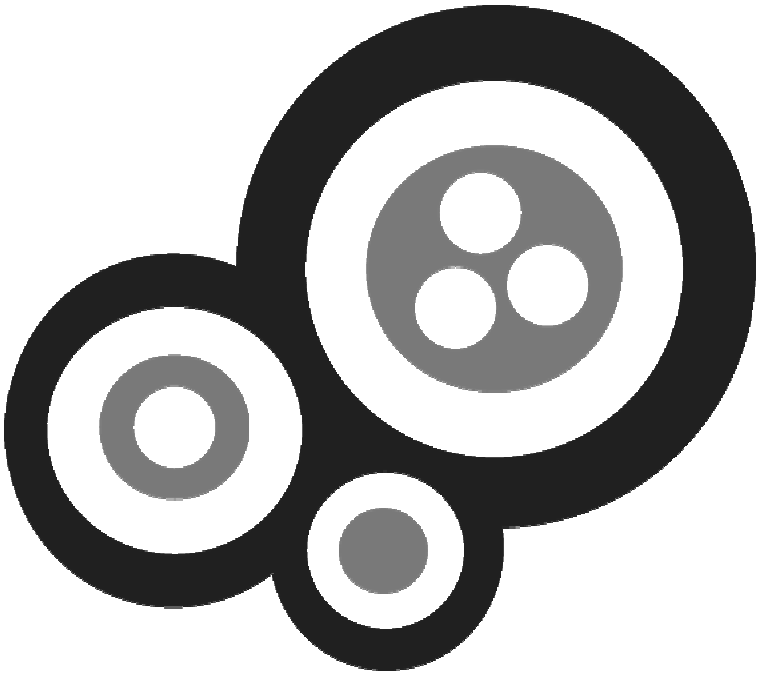

PLENARY 2A

PRECONCEPTION CARE FROM SCIENCE TO PRACTICE: INFORMATION FOR ACTION

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GENETIC RISK ASSESSMENT IN PRECONCEPTION CARE

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Basically there are two complementary methods for genetic risk assessment: History taking, followed by further work up if needed; and genetic screening.

History taking addresses existing indications of increased risk of inherited conditions, as well as risk factors for new mutations. Indications of increased risk of inherited conditions are disorders, present and past, in the couple, their previous children if any or extended family; consanguinity; ethnic background or community of origin. Risk factors for new mutations are age and exposure to mutagenic substances or ionizing radiation. Taking a family history involves drawing a pedigree, including at least the grandparents of the couple and all their descendants. For each person in the pedigree one has to ask which disorders they have now or have experienced in the past. Just asking whether there are any inherited diseases or congenital disorders in the family is not sufficient. Once an indication for the possible existence of an inherited disorder in the couple or family is found, one may first try to document the evidence and consult an expert or refer to a genetic clinic right away.

Genetic screening is in some places available for the detection of carriers of recessive diseases such as cystic fibrosis, sickle cell disease, thalassemia, or other disorders with a high prevalence in the population or locally. As sensitivity of mutation detection may be less than a hundred percent, screening should be restricted to couples without a family history of the disease at issue. In case of a positive family history starting with determining the mutation in the index case followed by a cascade approach, is better.

When a couple turns out to be at risk for a genetic disorder in their child, genetic counseling including a review of existing reproductive options is indicated. Genetic risk assessment in preconception care requires making a clear distinction between advice that is aimed at modifying behaviour in cases where risk factors themselves can be influenced and non-directive information aimed at increasing reproductive autonomy where they cannot.

PRECONCEPTION HEALTH PROMOTION MEANS AND CONSTRAINTS ANALYSIS

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Background: (1) There is no organized preconception care in Belgium but only opportunity care, with great variability in attitudes. (2) The incidence of neural tube defects is above the expected rate. (3) The percentage of pregnant women with correct folates intake in diet (400 µg per day) is near 0%. (4) The percentage of women taking folates supplementation before pregnancy is only 15%. (5) The percentage of women who consult before pregnancy is very low.

For these reasons, ONE (Office for Birth and Childhood) has proposed axes to promote preconception health.

Methodology: These strategies are: (1) organization of a campaign to sensitize population and health care providers; (2) edition and presentation at scientific meetings of guidelines for primary care providers and organizations involved in maternal and childhood health and (3) implementation of a pilot preconception clinic.

Results: 1. Among means proposed to promote preconception care, (i) posters and leaflets distributed in primary care services seem to be more efficient than media; especially short TV spots; (ii) the implementation of a pilot clinic seems to enhance all activities, outside the clinic, in the field of preconception health.

2. Among constraints, we observed (i) a high level of unplanned pregnancies, and thus a low level of women benefiting preconception care; (ii) the difficulty to provide immunization, due to the necessity to postpone a pregnancy after assessment of the immune status and possible immunization process; (iii) the poor compliance of women to take folates for a long period without pregnancy; (iv) the poor compliance of providers to the guidelines; (v) the absence of training of providers about the ethical stakes associated with management of genetic disorders ; (vi) the difficulty for many practitioners to integrate the concept of democratic eugenics in their practice; (vii) and to assess the stochastic dimension of clinical manifestations of inherited diseases.

Conclusions: From this experience, we propose (1) the implementation of a folates fortification program to avoid the risks associated with uncertain preconception visit; (2) the relay of immunization by scholar medicine and (3) a training of providers about ethical issues of preconception care, especially for genetic disorders.

WHAT ARE THE CHALLENGES AND KNOWLEDGE GAPS FOR IMPLEMENTING PRECONCEPTIONAL HEALTH?

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Lack of progress in adopting preconceptional health as a standard of care for all women of childbearing potential can be explained by several attitudes and circumstances (Moos, 2004) including:

- Assumption that women are already getting the care they need
- Insufficient education of clinicians about preconceptional health
- Lack of confidence that preconceptional health counseling will make a difference
- Belief that women will seek care appropriate to their needs
- Inadequate appreciation that many pregnancies are unintended
- Concerns about the costs of providing the service
- The existing paradigm of prenatal care as the favored prevention strategy for poor pregnancy outcomes is entrenched

What is needed is a research agenda to move science to practice—important research questions include:

- What interventions are most likely to impact clinicians' knowledge, attitudes and skills around preconceptional health?
- What are the most effective strategies for engaging clinicians in considering the woman's reproductive life course at every encounter?
- What incentives are necessary to engage clinicians to assessing and addressing a woman's health beyond the stated purpose of her visit (e.g. assess family planning desires and needs for women who present for the care of a chronic disease that is known to impact the woman, fetus or infant if pregnancy occurs without special disease management)?
- How do we encourage and monitor the delivery of evidence-based care in the arenas of preconceptional health promotion and disease prevention?
- What are the most effective strategies to alter lifestyle and other risks prior to conception to positively impact women's risks to future pregnancies?
- What clinical and community-based strategies are proven to impact the intendedness rate?
- What are the most efficacious vehicles for women receiving and acting on preconception health messages?
- What are the benefits of introducing reproductive life plans into the care of all men and women?
- What are unintended consequences of the preconception health agenda?
- What strategies are most successful in changing an entrenched (but largely ineffective) prevention model?