

## **PUBLIC CONSULTATION RARE DISEASES: EUROPE'S CHALLENGES**

### **Submission on behalf of the *EurOSS* initiative**

The *EurOSS* initiative comprises a group of clinicians and researchers from across Europe committed to raising awareness about rare obstetric diseases and events, their prevention and optimal management through Europe-wide research and its implementation in practice.

#### **Preamble**

At least 90% of the female adult population of Europe becomes pregnant at least once during her reproductive lifetime. Thus, nearly half of the entire population of Europe will be at risk of both the common and the rare diseases and events associated with pregnancy, labour and delivery at least once. Despite the potential risk faced by many millions of women rare obstetric diseases and events are not specifically listed as one of the important categories of conditions in the rare diseases consultation and we believe this is a grave oversight.

Rare obstetric diseases and events include conditions which are specific to the pregnant state (eg. eclampsia, post-partum haemorrhage requiring significant intervention and amniotic fluid embolism) and conditions which may present outside of pregnancy but to which the pregnant state adds further complications (eg. maternal pulmonary vascular disease and maternal heart disease (both congenital and acquired)). Many rare obstetric conditions can be characterised as "near-miss" events and most are thought to present a greater burden of disease than maternal mortality statistics reflect. Rare obstetric diseases and events cannot be always be anticipated ahead of their occurrence and can rarely be 'tested' for ahead of the event.

In keeping with many other rare diseases, rare obstetric diseases and events are, as a consequence of their rarity, under-researched and their management is seldom evidence-based. The vast majority of rare obstetric diseases and events can be best characterised by a lack of evidence as to their incidence, their risk factors and pathophysiology, and, with the notable exception of eclampsia, by the lack of an evidence-base as to the most effective modality of management.

#### **Consultation questions**

##### **1. Is the current EU definition of a rare disease satisfactory?**

The *EurOSS* initiative agrees that the current definition is satisfactory. This definition been used by the only currently operating population-based system designed to investigate rare obstetric diseases and events.<sup>1,2,3</sup>

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<sup>1</sup> Knight M, Kurinczuk JJ, Tuffnell D, Brocklehurst P. The UK Obstetric Surveillance System for rare disorders of pregnancy. *Br J Obstet Gynaecol* 2005; 112: 263-265.

<sup>2</sup> <http://www.npeu.ox.ac.uk/ukoss>

<sup>3</sup> Zwart JJ, Richters JM, Öry F, de Vries JIP, Bloemenkamp KWM, van Roosmalen J. Severe maternal morbidity in the Netherlands: a nationwide population based cohort study of 371.000 pregnancies. *Br J Obstet Gynaecol* 2008; in press.

**2. Do you agree that there is a pressing need to improve coding and classification in this area?**

The *EurOSS* initiative agrees that this is a vital need. Ensuring consistency of classification and coding is an essential stage in any pan-European disease initiative. It is vitally so for rare obstetric diseases and events, many of which are diagnosed on the basis of clinical signs and symptoms rather than physiological or genetic testing which can be more easily standardised.

**6. What can be done to further improve access to quality testing for RD?**

We agreed that access to quality testing for RD is a vital aim. However, it must not be overlooked that many rare diseases in the obstetric field cannot be tested for *a priori*. Nevertheless, many women face the risk of rare obstetric diseases and events and the need to prioritise research in this area must not be ignored.

**14. Do you consider it necessary to establish a new European Agency on RD and to launch a feasibility study in 2009?**

We agree that rare diseases need to be highlighted and that a new European Agency devoted to this work will raise the profile of rare diseases. Through this route it will be important to ensure, that rare diseases such as those affecting women during pregnancy, labour and delivery are not forgotten since they have the potential to affect millions of women across Europe each year.

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