Office for International Relations and Mobility
University of Brescia, Italy

Karolinska Institutet

International Seminar:

Role of genes coding for cellular transporters in the toxicity of environmental pollutants: the example of SLC30A10

April 18, 2017
Aula Consiliare, School of Medicine
14:00-15:00

By Karin Broberg, Professor in Environmental medicine, genetics and epigenetics,
Institute of Environmental Medicine, Metals and Health, Karolinska Institutet, Stockholm, Sweden;
Senior Lecturer in Occupational and Environmental Medicine, Lund University, Lund, Sweden

Seminar's topic: Environmental toxicants and pollutants adversely affect human health, while genetic factors increase individual susceptibility. Identification of this predisposition can inform our understanding of the mechanisms of toxicity. To identify mechanisms of tolerance and susceptibility, Prof. Karin Broberg has screened a large number of genetic variants in relation to biomarkers of exposure and toxicity in human populations. Her research demonstrates that polymorphisms in genes encoding enzymes involved in the metabolism of chemicals serve as a protective mechanism from toxicity, and recently she showed the first example of adaptation to a toxic environment in humans. By using whole genome expression and epigenome data, she has linked genetic effects to alterations of gene expression epigenetics related to arsenic, cadmium, mercury, and manganese (Mn) exposure.

Manganese is an essential element that can become neurotoxic with a narrow range between essential and toxic doses. High Mn levels can be caused by industrial contamination, such around Brescia, or naturally occurring high levels in food and drinking water. Variation in genes coding for Mn transporters can also cause neurological symptoms. The *SLC30A10* cell surface protein is a Mn efflux transporter, and a rare *SLC30A10* loss-of-function mutations leads to severely elevated Mn concentrations and neurodegenerative symptoms. More frequent genetic variations of SLC30A10 were associated with neurological performance in children and adults from Brescia, and strongly contributed to variations in Mn biomarkers indicating a potential influence on neurodevelopment via Mn homeostasis.

Registration: Prof. Donatella Placidi donatella.placidi@unibs.it
Information: Prof. Roberto Lucchini roberto.lucchini@unibs.it