

# DBMR Research Conference

Seminar Room EG050  
Murtenstrasse 24, 3008 Bern

Date **Monday, 5 September 2022, 17:00 – 18:00**

Title **Blurring the boundaries of common and rare diseases**

Speaker **Prof. Alexandre Reymond, Ph.D.**  
**Director Center for Integrative Genomics, University of Lausanne**

**Biosketch:** Alexandre Reymond carried out his thesis in the laboratory of Dr. Viesturs Simanis at the Swiss Institute for Experimental Cancer Research (ISREC) and received his Ph.D. from the University of Lausanne in 1993. After completion of his postdoctoral training with Dr Roger Brent in the Department of Molecular Biology, Massachusetts General Hospital and in the Department of Genetics, Harvard Medical School in Boston, he moved to the Telethon Institute of Genetics and Medicine (TIGEM) in Milan in 1998 to lead a research group. He joined in 2000 the Department of Genetic Medicine and Development, University of Geneva Medical School. He moved to the Center for Integrative Genomics in October 2004 and became its Director in 2015. He was the President of the European Society of Human Genetics from 2019 to 2021.

**Abstract:** The recurrent 600 kbp deletions and duplications at human chromosome 16p11.2 are among the most frequent genetic causes of neurodevelopmental and psychiatric disorders, as they are found in 1% of individuals with autism spectrum disorders and schizophrenia. These rearrangements cause reciprocal defects in head size, body weight and as described below age of menarche (AaM). These rearrangements are mediated by human-specific duplications that appeared at the beginning of the modern human lineage, suggesting that their expansion has a possible evolutionary advantage that outweighs the accompanying chromosomal instability. These duplications include BOLA2, a gene involved in the maturation of cytosolic iron-sulfur proteins. The expansion of BOLA2 might have evolved to protect humans against iron deficiency as our species successfully expanded its ecological range at the cost of increased predisposition to rearrangements associated with autism.

**Prof. Alexandre Reymond has been invited by the Stem Cell Research and Regenerative Medicine (SCRM) Platform Bern.**

The DBMR cordially invite all colleagues and interested people to attend the Research Conference. An apéro will follow the Research Conference.

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Thursday, October 13, 2022

Next DBMR Research Conference

Rahul N Kanadia, Ph.D., University of Connecticut  
*Title: tba*



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