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PhD in Medical Sciences
2015-2016

INVITATION to the Public defence of

Kathelijn KEYMOLEN

To obtain the academic degree of '**DOCTOR IN MEDICAL SCIENCES**'

Reproductive counselling in translocation carriers: new answers to an old question?**Tuesday 7 June 2016**

Auditorium **Vanden Driessche**, 17:00

Faculty of Medicine and Pharmacy, Laarbeeklaan 103, 1090 Brussel

How to reach the campus Jette:

<http://www.vub.ac.be/english/infoabout/campuses>



Vrije Universiteit Brussel

Summary of the dissertation

Chromosomes are the carriers and transporters of most of our genetic material. In most of us, 46 chromosomes per cell are present and these chromosomes have a well-defined structure. Some people have a different number of chromosomes in their cells -called numerical anomalies- whereas in others some chromosomes have a different structure, called structural anomalies. Robertsonian and reciprocal translocations are examples of structural chromosomal aberrations and they occur in the general population with a frequency of 1/1000 to 1/700. However, they are diagnosed more frequently in persons with recurrent miscarriages or reduced fertility. Couples in which one of the partners carries a balanced translocation often seek genetic advice with respect to their reproductive chances and options. The contacts with these couples at the Centre for Medical Genetics triggered the interest towards the reported research.

We retrospectively looked at the outcomes of Preimplantation Genetic Diagnosis (PGD) and prenatal diagnosis in couples carrying a Robertsonian or reciprocal translocation. We observed that PGD is a valuable option for these couples, especially for the ones suffering from infertility. On the other hand, prenatal diagnosis in ongoing pregnancies without PGD, showed normal/ balanced chromosomal results in the majority of the cases.

Comparison of the characteristics and the results in all groups, made us conclude that several factors have to be taken into account when counselling these couples. The type of translocation, the gender of the carrier as well as the personal and familial reproductive history are important elements to consider. However, reproductive counselling is more than a sum of medical facts and the final decision stays with the couple.

Curriculum Vitae

Kathelijn Keymolen was born in Asse on 10th of November 1967. She attended the Koninklijk Atheneum Asse and graduated summa cum laude from Medical School at the VUB in 1992. She was trained as a paediatrician in the Sint Pieter and UZ Brussel hospitals in Brussels. During her training in paediatrics, she spent one year in the Centre for Medical Genetics UZ Brussels. Afterwards she applied and obtained a grant from the Horlait d'Apsens foundation to be trained in clinical dysmorphology at the University of Liège with Professor Alain Verloes and at the Catholic University of Leuven with the team of Professor Jean-Pierre Frysns.

In 1999 she returned at the Centre for Medical Genetics UZ Brussels where she has been working ever since as a clinical geneticist. She is a member of the Belgian College for Human Genetics and chair person of the Dysmorphology working group of the Belgian Society of Human Genetics.