IN MEMORIAM

Eric Engel
Geneva, 12 October 1925 - Geneva, 9 September 2011

My feelings of sorrow are associated with the loss of a great person who was committed to his institutional tasks and to his patients, a scientist who influenced my historical and perspective vision of the field of human genetics and gifted me with friendly competent and inspiring conversations on art and humanities. Our thick correspondence continued till the 5th of August 2011, date of the last letter I received from him, which revealed a trembling calligraphy but a lucid analysis of the inexorable evolution of his disease.

Fig. 1 A sketch of E. Engel to outline the “heterochromatid kiss” underlying segmental UPD

Eric Engel was born in Geneva on 12 October 1925. He was educated at the Medical School of Geneva where he earned his medical degree in 1951 and received residence training in endocrinology at the Geneva University Hospital (1951-1960). In 1960, following a visit to the laboratory of Dr. J. Lejeune in Paris, he accepted positions as an instructor in Medicine at the Harvard Medical School and as a clinical and research Fellow at the Massachusetts General Hospital, where he distinguished himself in the field of cytogenetics. In the period 1960-63 he set up a laboratory for the study of chromosomes, shifting the approach from fibroblasts to short term blood cultures. His main interest focussed on sex chromosome anomalies of endocrine patients, who provided him with a unique opportunity of studying the first described cases of Klinefelter and Turner syndromes.

In 1963 one of the world's leading endocrinologists, G.W. Liddle recruited him to the Vanderbilt University School of Medicine to help establish a medical genetics unit and direct the cytogenetics lab. Dr. Engel was named professor of Medicine, associate
professor of Pediatrics and director of the Division of Genetics. During the long and very active period at Vanderbilt (1963-1978) he promoted the development of genetics, addressed the study of chromosomal changes in both constitutional pathology (birth defects and congenital disorders) and hematological malignancies (chronic myeloid leukemia, promyelocytic leukemia), and contributed to the mapping of human genes by the novel tool of somatic cell hybrids. By the early 1970s he had developed a reputation as one of the few leading cytogeneticists in the country.

Despite being a recognized leader of basic science research projects he always maintained his initial preference for clinical experience, nurtured by the constant practice of genetic counselling. As Director of the Genetics Center he worked on the problems of infertility and recurrent abortions in the first trimester. It was the high rate of chromosomal aberrations in early pregnancy losses that stimulated his first reflections on the potential for complementation of gametes that are nullisomic and trisomic for the same chromosome. This seminal breakthrough, which can be traced back to his last years at Vanderbilt University, was the first conceptual framework for thinking about Uniparental Disomy, the presence in a diploid genome of a chromosome pair derived from one parent. The concept was subsequently extended to include two alternative mechanisms, trisomy and monosomy rescue through chromosome loss or duplication following maternal non disjunction events, which accounted for UPD in the two forms of heterodisomy and isodisomy.

In 1979 Eric Engel moved back to Geneva to direct the University Institute of Medical Genetics. He further elaborated on the concept of uniparental disomy with its attendant risk of recessive disorders, a risk later compounded by the occurrence of imprinting, keeping up with all the new published evidence and critically reviewing the enormous body of data contributed from all over the world. He retired in 1991 and was succeeded by S.E. Antonarakis. He continued to be involved in science, and to be a prolific writer. His activity was reflected in numerous reviews and lectures and the world renowned book written together with Stylanos Antonarakis on “Genomic Imprinting and Uniparental Disomy in Medicine”.

The name of Eric Engel is indissolubly linked to the seminal concept of UPD. The serendipitous inspiration of this concept was a result of cross-fertilization of clinical experience with basic research interests in the field of constitutional chromosomal aberrations. The impact of this mechanism, at the root of human pathology, on prenatal and postnatal diagnostics has been incredible. This mechanism is essential for the understanding of genomic imprinting and the mechanisms associated with tumorigenesis and tumor progression.

Eric was not only a talented scientist, he was also a person of exceptional culture, a “connaisseur” of arts and a cordial and friendly interlocutor.

Thank you Eric for the gifts you have given us in Human and Medical Genetics and beyond!

Lidia Larizza
Medical Genetics
San Paolo Medical School
University of Milan
Via A. di Rudini 8, 20142 Milano, Italy