



## Stylios E. Antonarakis

Professor of Medical Genetics, Division of Medical Genetics, University of Geneva Medical School, Geneva Switzerland.

Director, Division of Medical Genetics, University and Cantonal Hospital of Geneva.

### CV

#### EDUCATION

Athens University School of Medicine, 1969-1975  
M.D. degree (magna cum laude) 1975  
Athens University School of Medicine  
Doctoral Thesis (magna cum laude) 1983

#### INTERNSHIP AND RESIDENCY

Internal Medicine, 1976-1978  
King Paul's University Hospital, Dept. of Medicine (Prof. G. Daikos)  
Hellenic Air Forces Gen. Hospital, Dept. of Medicine (Dr. G. Psimenos)  
Pediatrics, 1978-1980  
Kozani General Hospital, Dept. of Pediatrics (Dr. P. Economopoulos)  
Patras Children's Hospital, Dept. of Pediatrics (Prof. Th. Giogarakis)

Aghia Sophia University Children's Hospital, Dept. of Pediatrics (Prof. N. Matsaniotis)

#### LICENSES TO PRACTICE MEDICINE

Athens, Greece 1975  
State of Maryland, USA, 1984 License # D304  
Post-Doctoral Fellowship  
The Johns Hopkins University School of Medicine ;  
Department of Pediatrics, Genetics Unit (Jul 1980 - Mar 1983)  
Professor Haig H. Kazazian, Jr., M.D

#### CONTACT INFORMATION

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## Chromosome 21; a Small Genomic Land of Fascinating Disorders

Chromosome 21 is the smallest human chromosome, three copies of which are associated with Down syndrome. The determination of the nucleotide sequence of 33.5 Mb of DNA of the (almost) entire long arm was achieved after an international collaborative effort in 2000. The total number of genes of chromosome 21 has not yet been accurately determined, but the current estimate is approximately 240. In addition, a considerable sequence variation has been determined.

These achievements now provides unprecedented opportunities to understand the molecular pathophysiology of trisomy 21, elucidate the mechanisms of all monogenic disorders of chromosome 21, and discover functional sequence variations that predispose to common complex disorders. All of that requires the functional analysis of gene products and the determination of the sequence variation of this chromosome.