EDITORIAL

This volume focuses on different congenital malformations and analyses their genetic basis.

In the last 20 years a thorough knowledge on the molecular basis and on the biological mechanism involved in genetic illnesses, in partnership with the development of sophisticated technologies, has deeply modified both the genetic consultation and the diagnostic approach to these pathologies.

It was only 1975 when Edwin Southern developed the Southern blot and the first DNA test was then undertaken in a laboratory. This discovery led few years later to the first prenatal molecular genetic test that was performed in a case of hemoglobinopathy.

Then in 1985 the polymerase chain reaction was firstly described and allowed to execute diagnostic test in a timely fashion and from small biological samples.

Contemporarily the advancements on the human genome project have led to new knowledge on the molecular basis of genetic illnesses such as the imprinting and the anticipation phenomenon, the presence of micro deletions, of uniparental isodisomy, of gene instability and others.

Moreover new sophisticated technologies such as molecular cytogenetics and nanotechnology have been developed.

All these new acquisitions have opened new perspectives mainly in the field of prevention; genetic screening has been implemented and the geneticist consultation has become an essential turning point in the management of these type of patients.

During a genetic consultation not only genealogical information is collected and then the risk of recurrence calculated, but once considered the possible ethiopathogenesis, the genetic heterogeneity, the possible tests, a proper procedure is undertaken to reach a definitive diagnosis. As these are rare pathologies the consultation has also the importance of identifying a center that can perform the correct test and then proceed to a proper follow-up of the disease. These centers can be either national or international and nowadays are based on a multidisciplinary approach, therefore usually equipped with different professional figures. Nowadays all this knowledge can be used to face new challenges: multifactorial diseases might benefit from predictive genetic testing, moreover they might help in exploring the effect of inter-individual genetic differences on the pharmacokinetics, pharmacodynamics, efficacy, and safety of drug treatments.

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