# *"Standardization of molecular biology techniques for Down Syndrome diagnosis in amniotic fluid in Guayaquil-Ecuador: The intiation of a new era"*

Down syndrome (DS), the most frequent cause of mental retardation, caused by a proven chromosomal aberration, is characterized by a well defined and distinctive phenotype and natural history. It is caused by the triplication of all or a critical portion of the chromosome 21 also denominated for this reason trisomy 21 (1,2). It has been recognized that the risk of having an affected offspring increases with maternal age (3). Above 30 years the risk is 1 in 1,000 and above 40: 9 in 1,000 (4,5). Affected individuals most frequently have specific congenital malformations in the heart (30 to 40% in some studies), in the digestive system: duodenal stenosis or atresia, imperforated anus and Hirshprung's disease and are 10 to 20 times at higher risk than the normal population to develop leukemias and leukemoid reactions (6,7).

More than 30,000 deliveries are annually attended at the Enrique C. Sotomayor Obstetrics and Gynecology Hospital of Guayaquil-Ecuador. This is one of four health care providing facilities managed by the "Junta de Beneficencia de Guayaquil" a private non profit organization whose mission is to provide partially subsidized services in healthcare and education basically to the low socio-economic population of all ages of Guayaquil (8). In one study regarding adequacy of prenatal care it was reported that 18% of woman delivering at our institution are > 30 years of age (9), therefore 6,000 pregnant women per year are at higher risk of having affected offsprings. Due to technical and implicated costs prenatal diagnosis in guayaquil and in our institution has not been feasible. The Catholic University maintains an academic agreement with the "Junta de Beneficencia" by which a great number of postgraduate residency programs are performed. Up to date much of the research work performed at the Biomedicine Institute of the Catholic University has been carried out in cooperation with the teaching hospitals of the Junta Organization.

As a preliminary step our Institute carried a project with University funding by which chariotyping in peripheral blood was standardized (10). Despite this, cultivating cells from peripheral blood for cytogenetic diagnosis is tedious, depends on the investigator's experience and results are not rapidly obtained. In our case this step has only led us to neonatal diagnosis of chromosomal diseases. In order for prenatal diagnosis to initiate in Guayaquil we have foreseen step 2 and 3. Step 2 will commence this year with a small University funded project for the standardization of molecular biology techniques for Down Syndrome diagnosis in amniotic fluid mainly FISH and PCR techniques (11-14) in which results maybe obtained in 48 hours not only of trisomy 21 yet of 13 and 18. Our initiation will only be carried out among consenting term women in which cesarean section are to be performed from which amniotic fluid will be sampled aided us to standardize techniques. Meanwhile we will start an intensive sonographic invasive program commencing only with amniocentesis. After step 2 and 3 are accomplished we may begin prenatal diagnosis and genetic counseling in Guayaquil and be able to participate in future research collaboration with other developed world Universities.

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