Methods: DNA samples of 56 Egyptian and 17 Lebanese patients were screened for the CFTR gene mutations. The 27 exons and their flanking regions of the CFTR gene were amplified by PCR using the published primer pairs and were studied by automated direct DNA sequencing to identify disease-causing mutations.
Results: CFTR screening revealed the identification of seven mutations: including two novel (c.3718-24G>A; c.2782G>C) and five previously reported mutations (c.1418delG, c.2997-3000delAATT, c.902 A>G, c.2620-15C>G, c.3877G>A). Furthermore, six polymorphisms were found: c.1408A>G, c.3870A>G, c.2562T>G, c.1584G>A, c.4389G>A, c.869+11C>T. These mutations and polymorphisms were not previously detected in the Egyptian population except for the c.1408A>G polymorphism. Whereas Lebanese patients have a complex allele c.[869+11C>T;3909C>G] not previously described, no other novel mutations were identified.
Conclusion: Identification of CFTR mutations will become increasingly important in undocumented populations. The current findings will help us to establish a panel of the CFTR gene mutations in the Egyptian and Lebanese populations for designing an appropriate strategy for future genetic diagnosis of CF.

J17.15
Is Elucigene CF30 Kit effective in detecting CFTR mutations on Algerian patients?

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Introduction: Cystic Fibrosis (CF) [MIM # 219700] is the most common autosomal recessive genetic disease, in Caucasian populations [1].
In Algeria, no information is available about the incidence of CF. In the Maghreb, there are few data about the molecular basis of CF, probably due to an under diagnosis.
This study evaluated the effectiveness of the ELUCIGENE CF30 Kit in a sample of Algerian CF patients.
Patients and methods: Subjects: Twenty four (24) unrelated CF patients were recruited from the Pneumology and Allergology Department of the specialized Hospital Center in Canastel Oran (Algeria).CF diagnosis was based on a clinical findings and repeated positive sweat chloride tests (>60 mmol/l).
DNA extraction and genotyping
- 10 ml of whole blood was collected from all participants in EDTA tubes.
- Genomic DNA was extracted using standard Salting Out procedure [2].
- Mutations were explored by the ELUCIGENE CF 30 Kit (Tepnel Diagnostics, Oxon, United Kingdom) which is base d on a PCR/ARMS technique.
In screening the twenty four CF patients (48 chromosomes) for 30 mutations available in ELUCIGENE CF30 kit, only five mutations were found:
- c.1521_1523delCTT (F508del).
- c.579+1G>T (711+1G>T).
- c.1624G>T (G542X).
- c.3909C>G (N1303K).
- c.1652G>A (G551D).
All the mutations found were validated.
„ The Elucigene CF30 detected 5 mutations in our sample, which is about 16.66%.
„ The detection rate seems low compared to European populations. **Conclusion:** The most interesting is to develop in collaboration with ELUCIGENE one specific Maghreb Kit and inclued mutations which frequency higher than 2%.

J17.16
Notification of death from cystic fibrosis in Brazil during 30 years from 1981 to 2010

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The aim of this work was to evaluate the notification of cystic fibrosis (CF) as primary cause of death in Brazil, from 1981 to 2010, and to compare with developed countries. The Brazilian data was obtained from SIM-Data/SUS and the American from the CDC WONDER. The Brazilian median age at death (MAD) was 3.9 years in 1981 and 12.4 years in 2010 for typical severe CF. In 1994, the Brazilian MAD was much lower (7.2y) than that (21y) of seven developed countries reported by Forgarty et al, 2000. We found no increase in MAD in Brazil from 1981 to 1985; from 1986 to 2010 it showed a threefold increase. From 1981 to 1990, there were few deaths over 25 years of age in Brazil, mostly concentrated in younger age groups. During the sample period there was an increase in deaths in higher age groups in Brazil, which may reflect better patient survival rates due to increased knowledge of the disease, with repercussions for its diagnosis and treatment. We also observed an increase in the reported cases of CF deaths per 100.000 inhabitants in Brazil over the years, possibly due to the better knowledge of the disease and consequently more accurate death notification, since this increase is not typical of a genetic disease. For the USA we observed a decrease in notified CF deaths, despite the population increase which may be a reflection of a smaller number of people born with CF, due to appropriate genetic counseling and prenatal diagnosis.

J17.17
SERPINA1 gene polymorphism frequency in clinically confirmed cystic fibrosis patients

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Background - *SERPINA1* gene is known to be one of cystic fibrosis (CF) modifier gene. Patients who carry the Z allele are at greater risk of developing severe liver disease. The aim of this study - to assess whether PIZ (rs28929474), PIS (rs17580) M1 ala/val (rs6647) or M2 (rs709932) alleles are associated with liver disease in patients with CF. Materials and methods - 61 clinically confirmed cystic fibrosis patients (enrolled 1998- 2013) and 61 control patients (without confirmed cystic fibrosis) were analysed for M, S and Z alleles in *SERPINA1* gene. The methods used were multiplex PCR and RFLP. Results - M2 allele was more common in CF patients comparing to the control group, (MAF affected=0.2449, MAF controls = 0.123, p=0.018, OR=2,314, CI95%=1,138-4.705). Analysing haplotype rs28929474-rs17580-rs6647-rs709932 frequency in the *SERPINA1* gene in patients and controls significant association was found in CF patients with haplotypes N-N-val-M2 (MAF affected=0.244, MAF controls=0.123, p=0.0198) and N-N-val-wt (MAF affected=0.516, MAF controls=0.647, p=0.051). Comparing CF patients with confirmed liver damage (n=6) and CF patients without it statistically significant association was not found. Conclusions - Haplotypes in the *SERPINA1* gene do not predispose cystic fibrosis patients to liver disease/damage. M2 allele and its containing haplotypes are more common in cystic fibrosis patients than in control group.

J17.18
Population data of 11 DNA markers from a sample taken from South Romania

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In this work, were typed in 100 unrelated individuals (sex ratio 60:40) from South Romania eight DNA markers. Aim was to analyze the genetic variability and to establish the relation between this region and other European populations. We establish Hardy-Weinberg equilibrium, gene diversity, genetic distances and tree topology by PHYLIP 3.68 package. Polymorphism's frequencies were similar to the mean frequencies calculated for the whole set of populations included in the study. The mean value of Ht was 0,201, and for Hs was 0,18. The most affiliated population with our lot are Italy, Spain, Poland, Germany, Greece and Turkey the most distant population are United Kingdom, Sweden, Croatia and Slovacia.

J17.19
A new variant of Ehlers-Danlos syndrome with inborn errors of mucopolysaccharide metabolism in the mother and son

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Background: Connective tissue dysplasias are characterized by the clinical polymorphism and genetic heterogeneity. Each new patient with Ehlers - Danlos syndrome, according to our observations is potentially a new variant of the syndrome.
Care report: Family V. has been followed-up for 13 years. Proband V. Clinical manifestations - coarse facial features, a disproportionate body, keeled chest deformity, kiphoscoliotic spinal deformity, wing-like scapulas, an increased skin extensibility, its softness, velvet, hypotonia, hypermobility of joints, varicose veins, swelling of the lower extremities. Proband's mother M. - disproportionate body, coarse facial features, kiphoscoliotic spinal deformity, a soft, doughy, hyperelastic skin, hypermobility of joints, varicose veins, lymphatic edema of the lower extremities.
The examination echografically revealed hepatosplenomegaly, metabolic, dysplastic changes in the kidneys, mitral valve prolapse, an additional chord of the left ventricle. Biochemically - signs of an increased collagen degrada-