P032

The dynamics of DNA diagnosis availability for cystic fibrosis patients in the Russian Federation, and genetic variation analysed using the National Disease Registry between 2013–2018

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Objectives: To analyse the dynamics of detecting genetic variants of CFT gene based on data from the Registry between 2013 and 2018 in the Russian Federation (RF).

Methods: Registry for 2013 year contained data of 1968 patients from 74 different regions, for 2018 year – 3142 patients from 81 RF regions.

Results: 94.3% out of all CF patients underwent genetic testing in 2018 compared to 87.5% in 2013. Children who underwent the testing composed 94.6%, adults – 93.6% of the cases in 2018 (compared to 87.5% for children and 91.0% for adults in 2013). The overall frequency of allele identification made up to 89.3% (89.1% in 2013). Out of the entire group of patients who underwent genetic testing, 82.4% had 2 variants of DNA sequence of the CFT gene (66.1% in 2013), 1% had only one variant, and in 3.7% none of the genetic variants was identified (in 2013, 25.9% had one variant, 4% had none).

A total of 212 genetic variants were revealed (112 in 2013). The most common genetic variants are F508del - 53.05%, CFTRdele2,3 - 6.09%, E92K - 3.04%, 3272-16T>A, D579Y and 3849+10kbC->T - 2.38%, 2143delT - 2.11%, 2184insA - 1.84%, 1677delTA - 1.58%, L138ins - 1.58%, R951X - 1.58%, 1959-12insC - 1.58%, and 1983delN - 1.58%. In the RF, the most common (in 2013, 25.9% had one, 8% had none). A total of 212 genetic variants were revealed (112 in 2013). The most common genetic variants are F508del - 53.05%, CFTRdele2,3 - 6.09%, E92K - 3.04%, 3272-16T>A, D579Y and 3849+10kbC->T - 2.38%, 2143delT - 2.11%, 2184insA - 1.84%, 1677delTA - 1.58%, L138ins - 1.58%, R951X - 1.58%, 1959-12insC - 1.58%, and 1983delN - 1.58%. In the RF, the most common (in 2013, 25.9% had one, 8% had none).

Conclusion: The L138ins is a rare variant CFT in the world, but it is often found in CF patients in the RF, to a greater extent in adult patients. The widespread introduction of DNA diagnostics, the inclusion of the L138ins in the panel of common variants in the CFTR, the detection of CF among adults has significantly increased the diagnosis of this variant in Russia. The L138ins was more common in CF patients living in the Middle Urals. We would like to thank CF Patient Registry of the RF for providing access to patient data and thank the individual regional CF centres’ representatives for allowing use of data (http://mukoviscidoz.org/).

P034

Prevalence of cystic fibrosis paediatric patients with p.Arg1162X mutation in southern Brazil: a migration flow outcome

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Methods and objectives: Here we present data of CF paediatric patients from a referral center in the South of Brazil with at least 1 allele of p. Arg1162X mutation. The objective of this study was to report the prevalence of this variant in Brazil and relate it to migration flows.

Results: In our database of 87 CF paediatric patients, p.Arg1162X is the second most common (4.4%) mutation, a higher prevalence than that in the whole country database (3.8%, p = 0.05). Seven of the patients have at least 1 allele and 1 of them is homozygous for this mutation. The mean current age of patients is 10y. The majority of diagnosis was made after newborn screening (58%). In the latest Brazilian CF Registry report, approximately 70% of CF patients live in the south and southeast regions, where the majority of European immigrants were settled in the past. The heterogeneity of Brazilian genetic background is principally influenced by immigration flows. Italian immigration to our region peaked in the 1880s and more than half of immigrants were from the Veneto region. Italian studies show that the pathogenic variant p.Arg1162X is very common in Veneto and Trentino regions (close to 10%).

Conclusion: Our higher prevalence of the p.Arg1162X mutation is probably due to a greater immigration of Veneto people to southern Brazil.

P035

Comparison of clinical findings of the patients with cystic fibrosis in terms of diagnosed with and without neonatal screening


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Cystic fibrosis in Cyprus: results from the national patients’ Registry

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Objectives: Since the early 1990s, systematic recording of cystic fibrosis (CF) patients’ data has been implemented in Cyprus. Recently, we have established and maintained a national patients’ registry. We aim to present a wide spectrum of genotypic and phenotypic features of CF patients in Cyprus from the most recent data collection in 2019.

Methods: Patients’ core data (demographics, diagnosis, genotype) and annual report data (growth, lung function, microbiology, complications, treatment) were systematically collected, recorded, and analysed following the specifications of the European patients’ registry online software system.

Results: Overall, data from 50 CF cases are presented, 5 of whom have deceased and 13 have been lost to follow-up in last years. Mean ± SD age at diagnosis was 6.2 ± 10.8 years, and mean ± SD age by the end of 2019 was 21.5 ± 14.3 years. Most commonly, patients presented at diagnosis with acute or persistent respiratory symptoms (46%), failure to thrive or malnutrition (42%), and dehydration or electrolyte imbalance (34%). In all cases, diagnosis was confirmed by genotyping. p.F508del was the most common mutation (45.2%), followed by p.Leu346Pro (6.7%), a mutation detected solely in individuals of Cypriot descent. According to the 2019 annual report, mean ± SD BMI-for-age z-score was 0.07 ± 1.3, whereas mean ± SD BMI % predicted was 78 ± 19.9. Haemophilus influenzae was the most common pathogen isolated in sputum cultures across all age groups (>60% of examined patients). Chronic colonisation with Pseudomonas aeruginosa and Staphylococcus aureus was confirmed in 37.5% and 50%, respectively, in patients who underwent a sufficient number of sputum cultures.

Conclusion: Systematic recording of patients’ data is necessary for the optimisation of healthcare for CF patients and for the overall improvement of disease prognosis.