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CEBPA gene mutations in Egyptian acute myeloid leukemia patients: Impact on prognosis

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Aim: To assess the prognostic role of myeloid transcription factor gene CEBPA (CCAAT/enhancer binding protein-α), a novel gene involved in leukemia in Egyptian adults AML.

Materials & Methods: Screening for CEBPA mutations was assessed using PCR-single-strand conformation polymorphism (PCR-SSCP) in pretreatment bone marrow samples from 55 newly diagnosed adult AML.

Results: CEBPA mutations were found in 11 (20%) of 55 AML patients. They had significantly higher hemoglobin (p=0.037) and lower LDH (p=0.003) levels when compared to those without. CEBPA mutations were frequently detected in M4 (45.5%) and M2 (27.2%) subtypes and significantly associated with normal karyotype (90.9%, p=0.007). We distinguished 6 cases with 2 different mutations or one homozygous mutation (CEBPA $^{double\text{-}mut}$) as well as 5 cases with only one single heterozygous mutation (CEBPA $^{single\text{-}mut}$). Patients with CEBPA mutations had significantly higher complete remission (p=0.047), lower mortality (p= 0.047). Double CEBPA mutant cases showed longer disease free survival (DFS) and overall survival (OS) when compared to wild type CEBPA (for DFS; median=27 versus 24 months respectively; p=0.009 and for OS; median=28 versus 25 months respectively; p=0.008). No significant differences were found between CEBPA $^{single\text{-}mut}$ cases and wild type cases regarding DFS and OS (for DFS; median=13 versus 24 months respectively; p=0.615 and for OS; median=14 versus 25 months respectively; p=0.703).

Conclusion: CEBPA mutation status is known to be a prognostic factor for favorable outcome in AML patients. CEBPA$^{double\text{-}mut}$ is associated with favorable DFS and OS. In contrast, CEBPA$^{single\text{-}mut}$ AMLs survival studies did not differ significantly with wild-type cases. These results demonstrate significant underlying heterogeneity within CEBPA mutation positive AML with prognostic relevance. Based on these findings, we propose that CEBPA$^{double\text{-}mut}$ should be clearly defined from CEBPA$^{single\text{-}mut}$ AML and considered as a separate entity in the classification of AML. Furthermore, incorporation of CEBPA mutation status into novel risk-adapted therapeutic strategies in Egypt will improve the currently disappointing cure rate of this group of patients.

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PPARγ and IL-6-174 G>C gene variants in Croatian patients with ischemic stroke

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Aim: Etiology of Ischemic Stroke (IS) is multi factorial and includes interaction of genetic and environmental factors. Different genes, their polymorphisms, host susceptibility and inflammation processes play a role in IS development. The aim of this study was to evaluate the effect of PPAR-γ and IL-6 gene variants on IS onset.

Material & Methods: A total of 301 subjects (144 male, 157 female) participated in the study, 114 patients with IS and 187 healthy controls.

Results: Statistically significant predictors of IS were gender (OR 7.13, 95% CI 2.92-17.39, p<0.001), hypertension (OR 7.82. 95% CI 2.53-24.19, p<0.001), lowered HDL cholesterol (OR 8.20, 95% CI 2.41-27.94, p=0.001), C-reactive protein (OR 5.26, 95% CI 1.92-14.41) and IL-6 -174 GC (OR 2.44 95% CI 1.01-5.91, p=0.0048) genotype. Males compared to females had 7 times higher odds for stroke. The presence of hypertension, lowered HDL and elevated C-reactive protein increased the risk of stroke. PPARγ was not statistically significantly associated with stroke. IL6-174 G/C genotype increased the odds for IS for 2.4 times.

Conclusion: We can point to the IL-6-174 G>C polymorphisms as candidate gene marker and risk factor for the prediction of ischemic stroke.

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