Summary

Pseudoexfoliation syndrome (PEX) is an age-related systemic disorder characterized by deposition of whitish-gray pseudoexfoliation fibrillogranular amyloid like material in several intraocular and extraocular tissues.The present study was carried out to investigate the association between*Tumor Necrosis Factor-alpha* and *lysyl oxidase like 1 (LOXL1)* genes variants with pseudoexfoliation syndrome in a group of Iraqi population.

This study was conducted on 45 patients (28 males and 17 females) with Pseudoexfoliation syndrome, their age ranges between (35 - 84)years, seen at Al-Diwaniya Teaching Hospital for the period from December 2014 to March 2015. Other 30 apparently healthy subjects (12 males and 18 females) were included as a control group. Blood samples were collected from both groups, genomic DNA was extracted from peripheral blood leukocytes for further molecular study to reveal any association between *Tumor Necrosis Factor-alpha*gene polymorphismat position -308 and single nucleotide (R141L) of *lysyl oxidase-like 1*gene polymorphisms and Pseudoexfoliation syndrome. Polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) technique was used for this purpose and digestion of the amplified DNA products by NcoI and AvaI endonuclease respectively.

The results of the study showed higher prevalence of pseudoexfoliation syndrome in the age group of 65-74 years, accounting for 64.36%, and 62.2% of the patients were males. The result also revealed that 55.6% of patients have pseudoexfoliation syndromewith cataract, 28.9% have pseudoexfoliation syndrome with cataract and glaucoma and 11.1% have pseudoexfoliation syndromewith glaucoma, while 4.4% of patients have

pseudoexfoliation syndrome only. The glaucoma distribution was 18 cases from 45 patients, 11 case was males while 7 case was females, and there was no significantassociation between unilateral and bilateral glaucoma. The data showed that 38 (84.4%) of patients have cataract, and the males showed a high tendency to have cataract than females.

Also, this study detected that the prevalence rate of single nucleotide polymorphism *Tumor necrosis factor-alpha* gene was not statistically significant among pseudoexfoliation syndromepatients,P=0.545. Heterozygous genotype GA and mutant A allele carrier frequencies not significantly in pseudoexfoliation syndromepatients P=0.808, P=0.601respectively, also homozygous normal GG genotype P=0.817; and the prevalence rate of single nucleotide (R141L) of *lysyl oxidase-like 1* genewas statistically significant among Pseudoexfoliation syndrome patients. The homozygous mutant genotype TT, P=0.0003, mutant allele T, P <0.0001, and heterozygous genotype GT was not statistically significant, P=0.091.

In conclusion,This study suggests that the *Tumor necrosis factor-alpha* gene -308 G/A polymorphism was not statistically significantrisk factors for the development pseudoexfoliation syndrome, while TT genotype and T allele of (R141L) of *lysyl oxidase-like 1* gene in patients with pseudoexfoliation syndromewere higher and significantly associated and may contribute to increase the progression of the pseudoexfoliation syndrome.