J Pharmacogenomics Pharmacoproteom 2018, Volume 9
DOI: 10.4172/2153-0645-C1-022



# Genetic polymorphisms in 665BP fragment of exon 10 in aryl hydrocarbon receptor related to acromegalic diseases

**Hasan F Alazzawie, Nathem M** and **Abbas M R Al-Khurasani**[2]
[1]Baghdad University, Iraq
[2]Al-Mustansiriya University, Iraq

A cross sectional biochemical and molecular genetics study was achieved from December 2016 to September 2017, enrolled 70 patients (36 males, and 34 females) with somatotrophic pituitary adenoma continuously attended to the national diabetic center in Baghdad. Their ages were between 20 to 70 years. This research aimed to observe the frequency of aryl hydrocarbon receptor (AHR) gene mutations and its association with growth hormone (GH), Insulin like growth factor-1 (IGF-1), and Insulin like growth factor binding protein-3 (IGFBP-3) concentration after more than one year of long acting octreotide treatment in samples of Iraqi acromegalic cases. Measuring the GH and IGF-1 concentrations were done using ELISA kits to find out the response of patient to the long acting octreotide injection. In addition measuring the IGFBP-3 was done to find out the relation with GH and IGF-1 and with the occurrence of the disease. Results observed that 20/70 (29%) of acromegalic (ACM) patients having high concentration of GH levels, 18/70 (26%) having high level of IGF-1 and 25/70 (36%) with high level of IGFBP3 concentrations, while other cases showed normal concentrations of GH, IGF-1 and IGFBP-3. Polymerase chain reaction (PCR) was used for the detection of *AHR* mutations as a cause of acromegaly, by using specific primers for amplification of exon 10 of *AHR* gene on chromosome no.7. Sequencing was applied for PCR products of *AHR* gene, ten different alterations in *AHR* gene were diagnosed. Four mutations in *AHR* were recognized in 20/70 (29%) patients which their codons A474T with substitution adenine by thymine (A/T), 176 and 180 with substitution thymine by adenine (T/A), and codon 411 with substitution of (C/T) for 665bp. Other two were recognized in 6/70 (9%) patients which are codon 705 with substitution (T/A) and codon 410 with substitution of (A/T) for 817bp. In addition another four recognized alteration were established in (22/70) (31%) which are codon 193 with substitution of (C/A), codon 344 with substitution of (T/A), codon 459 with substitution of (G/C), and codon 518 with substitution of (A/C) and this for 706bp of *AHR* exon 10. We concluded that ten novelties SNP were detected in Iraqi acromegalic cases.