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Research Article

Association of Vitamin D Receptor Gene SNP *BsmI* (1544410) with Axial Spondyloarthritis in Iraqi Patients Treated with Infliximab

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Abstract:

Background: Axial spondyloarthritis (AxSpA) is a chronic inflammatory disorder driven by tumor necrosis factor alpha and the interleukin-23/interleukin-17 pathway. Tumor necrosis factor alpha inhibitors such as infliximab improve outcomes, but responses vary, suggesting genetic influences; vitamin D receptor (VDR) polymorphisms—particularly *BsmI* (rs1544410) may modulate disease activity and therapeutic response.

Objectives: This study aimed to investigate the association between the VDR *BsmI* polymorphism and disease activity, as well as infliximab treatment response in patients with Axial spondyloarthritis.

Methods: A cross-sectional study was conducted on 150 AxSpA patients (108 males, 42 females) receiving infliximab for at least three months. VDR *BsmI* genotyping was performed using PCR-RFLP. The study took place at the Rheumatology Clinics of Baghdad Hospital, Medical City, Iraq.

Results: *BsmI* genotype distribution was AA (28%), AG (46%), and GG (26%), with allele frequencies of 51% (A) and 49% (G). No significant association was found between *BsmI* polymorphism and infliximab duration. However, a significant association was observed with disease activity. The AA and AG genotypes were also significantly associated with male gender. No significant correlations were found with other clinical features including inflammatory back pain, urinary tract infection, enthesitis, uveitis, dactylitis, psoriasis, or colitis.

Conclusion: The GG genotype was linked to an increased likelihood of inactive disease, while the AA genotype was associated with an increased risk of low disease activity. These findings suggest VDR *BsmI* polymorphism may influence AxSpA disease activity and response to infliximab

Keywords: Axial spondyloarthritis; Disease activity; Infliximab; Single nucleotide polymorphism; Tumor necrosis factor alpha; Vitamin D receptor.

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Introduction:

Spondyloarthritis (SpA) is characterized by inflammatory processes and new bone growth (1). A diagnosis of axSpA is often contemplated when persistent back pain manifests before the age of 45 years, however, cases of start beyond this age have also been documented (2). Axial Spondyloarthritis cases. Inflammation, loss of bone and cartilage, and subsequent remodelling with new bone formation occur in the entheses, axial skeleton, and peripheral joints. Patients with axSpA also exhibit various extra-articular manifestations, including inflammatory bowel lesions, psoriasis, and uveitis. Infliximab, a medication for AxSpA, is classified as a Tumor Necrosis Factor Alpha (TNF alpha) blocker that has profoundly altered the trajectory of inflammatory illnesses (3), including rheumatoid arthritis (RA), spondyloarthritis (SpA), and Crohn's disease (CD). Approximately 30% of patients do not exhibit a response to this therapy (4& 5). This absence of reaction may result from an initial non-response termed primary failure or from a decline in responsiv eness after an initial positive response referred to as secondary failure (6).

Immunogenicity has been implicated in instances of subsequent failure. It is characterized by the formation of antidrug antibodies (ADAbs) (7)(8). Research demonstrates a correlation between genetic VDR polymorphism and increased acute phase reactant levels as well as spinal bone mass density in individuals with axial spondyloarthritis (9). Numerous studies have examined the possible impact of VDR gene polymorphisms on susceptibility to AxSpA; nevertheless, the findings have been inconclusive(10). This study aimed to investigate the association between the VDR *BsmI* polymorphism and disease activity, as well as infliximab treatment response in patients with Axial spondyloarthritis.

Subjects and Methods

A cross-sectional study that included 150 participants, males (108) and females (42) with an age of ≥ 18 years, was conducted.

The study was conducted at the Rheumatology Clinics of Baghdad Teaching Hospital, Medical City, Baghdad, Iraq, and at the National Center Laboratories from May 2024 to September 2024. The

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protocol was approved by the Ethical Committee of the Department of Biochemistry and the College of Medicine, University of Baghdad, and by the Institutional Board of the Rheumatology Clinics of Baghdad Teaching Hospital, Medical City. All participants provided written informed consent, and all procedures adhered to relevant clinical biochemistry and rheumatology guidelines.

Inclusion criteria: were Adults (≥18 years) diagnosed with axial spondyloarthritis by a specialist rheumatologist using the Assessment of Spondyloarthritis International Society criteria (supported by magnetic resonance imaging or radiography), receiving infliximab for at least three months.

Exclusion criteria: Current use of anticonvulsants, antidepressants, or antipsychotics; other autoimmune disease; vitamin D or multivitamin supplementation; treatment with any tumor necrosis factor inhibitor other than infliximab within the last three months; pregnancy or lactation; and confirmed rheumatoid arthritis. Eight millilitres of blood were taken by vein puncture from patients and withdrawn after a 12-hour fast from all subjects. Two millilitres were isolated in an EDTA tube for genotyping, in addition to the following biochemical and serology tests (ESR). Serum was separated with blood centrifugation for 10 min and stored at −20 °C for biochemical analysis.

Genomic DNA was isolated from the whole blood of the patients. Appropriate primers are used for amplification of VDR genes to study the intended SNPs by using RFLP-PCR Method.

Patients who were included in this study have axSpA and on Infliximab treatment (biology treatment). All patients were interviewed, examined and diagnosed with axSpA depend on MRI or Xray that done at the Rheumatology Clinic of Baghdad Teaching Hospital, Medical City, Baghdad- Iraq. Patients included in this study were Male or female age more than 18 < years with Rheumatoid factor (RF) or anti cyclic citrullinated peptides (anti-CCP) positivity, baseline erythrocyte sedimentation rate (ESR) and C-reactive protein (CRP) levels.

Patients must take drug (infliximab) at last for 3 months according to criteria that classify adults with chronic back pain for at least three months if either of the following is met:

1. Imaging: sacroiliitis on imaging—defined as active inflammation on magnetic resonance imaging of the sacroiliac joints or definite radiographic sacroiliitis—plus at least one spondyloarthritis feature.
2. Clinical: human leukocyte antigen-B27 plus at least two spondyloarthritis features. Spondyloarthritis features include inflammatory back pain, peripheral arthritis, dactylitis, enthesitis (especially at the heel), family history of spondyloarthritis, good response to non-steroidal anti-inflammatory drugs, uveitis, psoriasis, Crohn's disease or ulcerative colitis, human leukocyte antigen-B27, and elevated C-reactive protein (11).

Statistical Analysis: Analysis of data was carried out using the Statistical Packages for Social Sciences-version 24. Data were presented in simple measures of mean, standard deviation, and range (minimum maximum values). The significance of differences of different means (quantitative data) was tested using Students t-test for differences between two independent means or the ANOVA test for difference among more than two independent means. Pearson correlation was calculated for the correlation between two quantitative variables, with its t-test for testing the significance of the correlation. Statistical significance was considered whenever the *P* value was equal to or less than 0.05.

Results

The study included 150 patients, mean±SD of age was 38.82 ± 8.88 years, and the youngest patient was 18 yrs. The oldest one was 62 years old. The mean ±SD of Body Mass Index was 28.92 ± 6.45 kg/m2. Patients were 28% are females and 72% were males. Only 16% of patients had a Family history of Axial spondyloarthritis, and 30% were smokers, as presented in Table 1.

BsmI polymorphism had 3 genotypes, AA 28%, AG in 46% and GG in 26% of patients. An allele frequency was 51% and the G allele frequency was 49% in Table 2.

Table 1: Demographic characteristics of participants

Variable			
Age (mean ±SD) (range) Years		38.82 ± 8.88	18-62
BMI (mean ±SD) (range) kg/m^2		28.92 ± 6.45	19.93 - 51.27
Gender No. , %	Female	42	28.0
	Male	108	72.0
Family of SPA No%		24	16.0
Smoking No., %		45	30.0
	Total	150	100.0

Table 2: Vitamin D receptor gene polymorphism *BsmI* variant distribution

	Polymorphism	No.	%
BsmI	AA	42	28.0
	AG	69	46.0
	GG	39	26.0
	Allele	No.	%
	A	153	51.0
	G	147	49.0

The *BsmI* genotype variant shows a statistically significant association with disease stage, $P \leq 0.008$. 62.5% of patients with very high disease activity had the AG genotype, 53.3% of patients with high disease activity had the AG genotype, compared to 75% of patients with inactive disease had the GG genotype. Patients with genotype GG had 10.8 times higher risk of having inactive disease, and patients with the AA genotype had 2.8 times higher risk of having low disease activity, as shown in Table 3.

Table 3: The association between the VDR polymorphism *BsmI* variant and disease stage

		Stages				Total	*P*-value
		inactive disease	low disease activity	high disease activity	very high activity		
BsmI	GG	9	15	9	6	39	0.008*
		75.0%	21.7%	20.0%	25.0%	26.0%	
	odd	10.8 (2.7-42.4)	0.65 (0.31-1.38)	0.62 (0.26-1.45)	0.93 (0.34-2.56)		
	p	0.0007	0.27	0.27	0.90		
	AA	0	27	12	3	42	
		0.0%	39.1%	26.7%	12.5%	28.0%	
	odd	/	2.82 (1.34-5.9)	0.9 (0.41-1.99)	0.32 (0.09-1.16)		
	p		0.005	0.8	0.08		
	AG	3	27	24	15	69	
		25.0%	39.1%	53.3%	62.5%	46.0%	
	odd	0.72 (0.18-2.86)	1.01(0.54-1.87)	1.5 (0.75-3.07)	2.1 (0.86-5.23)		
	p	0.64	0.97	0.23	0.09		

AA and AG genotypes of *BsmI* polymorphism were significantly associated with male gender, p≤ 0.01. The presence of UTI, psoriasis, and colitis were significantly not related to *BsmI* genotype, $P \leq 0.005$, 0.01 and 0.008, as presented in Table 4.

Table 4: Association between the genotypes of the *BsmI* polymorphism with gender and other disease manifestations

		BsmI			Total	*P*-value
		GG	AA	AG		
gender	Female	12	18	12	42	0.01
		30.8%	42.9%	17.4%	28.0%	
	Male	27	24	57	108	
		69.2%	57.1%	82.6%	72.0%	
Inflammatory back pain	No	9	9	21	39	0.51
		23.1%	21.4%	30.4%	26.0%	
	Yes	30	33	48	111	
		76.9%	78.6%	69.6%	74.0%	
Arthritis	No	15	15	27	57	0.91
		38.5%	35.7%	39.1%	38.0%	
	Yes	24	27	42	93	
		61.5%	64.3%	60.9%	62.0%	
UTI	No	27	39	48	114	0.005
		69.2%	92.9%	69.6%	76.0%	
	Yes	12	3	21	36	
		30.8%	7.1%	30.4%	24.0%	
Enthesitis	No	36	42	66	144	0.13
		92.3%	100.0%	95.7%	96.0%	
	Yes	3	0	3	6	
		7.7%	0.0%	4.3%	4.0%	
Uveitis	No	33	36	63	132	0.5
		84.6%	85.7%	91.3%	88.0%	
	Yes	6	6	6	18	
		15.4%	14.3%	8.7%	12.0%	
dactylitis	No	30	39	63	132	0.06
		76.9%	92.9%	91.3%	88.0%	
	Yes	9	3	6	18	
		23.1%	7.1%	8.7%	12.0%	
Psoriasis	No	36	42	69	147	0.01
		92.3%	100.0%	100.0%	98.0%	
	Yes	3	0	0	3	
		7.7%	0.0%	0.0%	2.0%	
colitis	No	39	42	63	144	0.008
		100.0%	100.0%	91.3%	96.0%	
	Yes	0	0	6	6	
		0.0%	0.0%	8.7%	4.0%	
Total		12	69	45	150	
		100.0%	100.0%	100.0%	100.0%	

Discussion

This cross-sectional study of 150 Iraqi patients with axial spondyloarthritis (axSpA) treated with infliximab shows that the *VDR BsmI* polymorphism (rs1544410) does **not** alter overall susceptibility to axSpA, yet it decisively influences therapeutic outcomes and disease activity. Mean BMI ≈ 29 kg/m²—overweight range; the manuscript does not report any correlation between BMI and BASDAI/ASDAS or infliximab response that disagreed with Queiro (12) that showed obesity is repeatedly linked to higher disease activity and ~20–30 % lower odds of achieving ASDAS-major improvement on TNF-inhibitors(13). 30 % of patients are current smokers, but it disagreed with (Queiro

et al., 2024) that showed smokers have higher CRP, slower BASDAI fall, and ~1.3× risk of persistent activity despite TNF. Mechanisms include excess oxidative stress and altered gut microbiota (14).

Genotype–activity link. Carriers of the GG genotype were over-represented among patients who attained an inactive disease state; whereas the AA genotype was enriched in those with low-to-moderate activity. These trends mirror a Spanish cohort in which GG homozygotes had the highest likelihood of clinical remission after anti-TNF therapy, reinforcing the idea that *BsmI* tracks pharmacodynamic response rather than baseline risk (15,16).

Sex-specific distribution. AA and AG genotypes were significantly more frequent in men. This pattern aligns with evidence that estrogen enhances *VDR* expression and vitamin-D-mediated anti-inflammatory effects, potentially moderating disease severity in females. In contrast, lower baseline *VDR* activity in males may amplify genotype-dependent variability after TNF inhibition (17,18). Parallel observations by Wielińska et al. (2024)—who found no *BsmI* effect on rheumatoid-arthritis susceptibility but a clear impact on TNF-inhibitor response—support a sex-influenced pharmacogenetic role for this variant (19,20).

Extra-articular features. No associations emerged between *BsmI* and uveitis, enthesitis, inflammatory back pain, or dactylitis, indicating limited influence on musculoskeletal or ocular sequelae. Weak negative correlations were noted for urinary-tract infection, psoriasis, and colitis (all $p < 0.01$), but the clinical weight of these findings appears minimal (20).

Duration of benefit. The polymorphism did not predict how long infliximab's effect persisted, implying that remission longevity may hinge on factors such as drug immunogenicity or vitamin-D status rather than genotype alone (21).

Biological rationale. Located in the 3′ UTR, *BsmI* may stabilize VDR mRNA, thereby elevating receptor abundance and potentiating 1,25-- $(OH)_2$-vitamin D-driven transcription of anti-inflammatory mediators (e.g., IL-10, CTLA-4). Enhanced *VDR* signalling could therefore complement TNF blockade, jointly curbing inflammation and aberrant bone formation (21).

Clinical implications Personalized medicine: *BsmI* genotyping can identify axSpA patients most likely to achieve remission on infliximab, guiding early biologic selection or escalation.
Risk stratification: Male carriers of AA/AG genotypes may warrant tighter monitoring because of their association with persistent activity.

Limitation

The study was conducted at a single Rheumatology Centre in Baghdad and included only Iraqi patients. This may limit the generalizability of the results to other populations with different ethnic or genetic backgrounds.

Lack of Vitamin D Level Assessment: Although the study investigated a polymorphism in the vitamin D receptor gene, it did not measure serum vitamin D levels. The absence of this biochemical data prevents the evaluation of potential interactions between genetic variation and vitamin D status.

Uncontrolled Confounding Variables: Potential confounding factors such as dietary habits, sun exposure, disease duration, infliximab, dose, physical activity and medication adherence were not controlled or evaluated, which may have influenced the observed associations.

small sample size for genotype subgroups: While the overall sample included 150 patients, the number of patients in each genotype subgroup (especially GG = 39, AA = 42) may be relatively small to detect subtle associations with extra-articular manifestations or secondary outcomes.

Conclusions

The study findings indicated a modulatory function of the VDR *BsmI* polymorphism in the clinical progression of axSpA during infliximab treatment, specifically affecting remission rates and gender disparities. Future multicentre studies that include vitamin D levels are necessary to validate these relationships and convert them into genotype-guided therapy protocols.

Authors' declaration

We confirm that all the Figures and Tables in the manuscript belong to the current study. Besides, the Figures and images, which do not belong to the current study, have been given permission for re-publication attached to the manuscript. Authors sign on ethical considerations' Approval-Ethical Clearance: The project was approved by the local ethical committee in (College of Medicine, University of Baghdad, Rheumatology centre in Baghdad, Medical City Complex, Baghdad, Iraq) according to the code number (M69) on (28/ 02/ 2024).

Conflict of Interest: None

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Data availability Upon reasonable request, the corresponding author will make the data sets generated and/or analysed during the current work available.

Authors' contributions

Study conception & design: (Manal K. Rasheed & Nizar AJassim). Literature search: (Abdullah H. Drewil .) Data acquisition: (Abdullah H. Drewil) Data analysis & interpretation: (Abdullah H. Drewil &Manal K. Rasheed). Manuscript preparation: (Abdullah H. Drewil , Manal K. Rasheed and Nizar AJassim). Manuscript editing & review: (Abdullah H. Drewil , Manal K. Rasheed and **Nizar AJassim**).

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علاقة الجين SNP *BsmI* (1544410) الخاص بمستقبلات فايتامين D مع التهاب اللاصق الفقاري في المرضى العراقيين المعالجين بعلاج انفلاكساماب

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المقدمة: يعد التهاب الفقار المحوري (AxSpA) اضطرابا التهابيا مزمنا تسهم في تسويغه كل من عامل نخر الورم ألفا (TNF-α) ومسار إنترلوكين-23/إنترلوكين-17. تحسن مثبطات عامل نخر الورم—مثل الإنفليكسيماب—المخرجات السريرية، غير أن تغاير الاستجابة بين المرضى يوحي بتأثيرات جينية محتملة؛ إذ قد تعدل تعددات أشكال جين مستقبل فيتامين د—(VDR) ولا سيما— BsmI (rs1544410) نشاط المرض والاستجابة العلاجية.

الأهداف: استقصاء العلاقة بين تعدد شكل VDR BsmI ونشاط المرض، وكذلك الاستجابة للعلاج بالإنفليكسيماب لدى مرضى التهاب الفقار المحوري.

الطرق: درست عينة مقطعية شملت 150 مريضا بـ AxSpA (108 ذكور، 42 إناث) يتلقون الإنفليكسيماب لمدة لا تقل عن ثلاثة أشهر. أجري تحديد النمط الجيني لتعدد شكل BsmI بتقنية تفاعل البوليميراز المتسلسل متبوعا بتعدد أطوال القطع الناتجة عن إنزيمات التقييد (PCR-RFLP). أجريت الدراسة في عيادات الروماتيزم بمستشفى بغداد، مدينة الطب، العراق.

النتائج: توزعت الأنماط الجينية على النحو الآتي(28%) AA :؛ و(46%) AG، و(26%) GG، وكانت تواترات الأليلات A = 51% :و G = 49%.لم يسجل ارتباط ذو دلالة إحصائية بين تعدد شكل BsmI ومدة العلاج الإنفليكسيماب. بالمقابل، ظهر ارتباط ذو دلالة مع نشاط المرض. كما ارتبط النمطان الجينيان AA و AGعلى نحو معنوي بكون المريض من الذكور. ولم تلاحظ ارتباطات معنوية مع السمات السريرية الأخرى، بما في ذلك ألم الظهر الالتهابي، والتهاب المسالك البولية، والتهاب الارتكاز، والتهاب القزحية، والتهاب الأصابع الشامل (الدكتيليتس)، والصداف، أو التهاب القولون.

الاستنتاج: ارتبط النمط الجيني GG بزيادة احتمال المرض غير النشط، في حين ارتبط النمط AA بارتفاع خطر النشاط المرضي المنخفض. تشير هذه النتائج إلى أن تعدد شكل VDR BsmI قد يؤثر في نشاط مرض التهاب الفقار المحوري وفي الاستجابة للإنفليكسيماب