

Title	Vitamin D receptor gene Bsml (rs1544410) polymorphism and radiological characteristics of Multiple Sclerosis
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INTRODUCTION

The pathophysiology of Multiple Sclerosis (MS) is complex and is probably responsible for the clinical and radiological heterogeneity of the disease. Vitamin D is one the environmental factors that plays a crucial immunomodulatory role and may impact the radiological features of MS.

AIM

This study aimed to investigate the association between vitamin D status, Vitamin D receptor (VDR) gene Bsml (rs1544410) polymorphism and radiological characteristics of MS.

METHODS

We conducted a cross-sectional study including MS patients followed at the Neurology Department of Charles Nicolle Hospital. Brain and spinal cord magnetic resonance imaging (MRI) was analyzed. Serum 25-hydroxyvitamin D levels were measured using chemiluminescence. The rs1544410 VDR gene polymorphism was studied for all patients.

RESULTS

73 patients. Sex ratio=0,4. Mean age: 40±11 years. Radiological characteristics: Brain atrophy on onset (13%), infratentorial lesions (71%), spinal cord involvement (66%), contrast enhancing lesions (42%). Vitamin D level was measured in 27 patients. 67% of patients had low vitamin D level. We didn't found significant statistical associations between vitamin D level and MRI data.

RESULTS

In the genetic study, the CC genotype and C allele were identified as the wild-type genotype and allele. We didn't found significant statistical associations between genotypes and lesions load distribution. Cerebral atrophy at onset was more frequently found for the CC genotype (p=0,02) and C allele (p=0,02) (table 1).

DISCUSSION

We observed that the C allele and the CC genotype were significantly associated with the presence of brain atrophy on the initial MRI. These findings suggest a predominantly degenerative disease profile associated with this genotype. On the other side , according to the literature, a higher levels of vitamin D have been associated with fewer signs of disease activity (presence of new lesions), but no relationship has been found with brain volume loss [1]. An interventional study by Dörr et al. examined the effect of low versus high-dose vitamin D3 supplementation on the number of new lesions, contrast enhancement, and brain atrophy, and found no significant results [2].

CONCLUSION

Vitamin D and its receptor are strongly involved in the pathophysiology of multiple sclerosis, and vitamin D deficiency may influence the disease course . Our results may represent preliminary findings and could encourage further prospective observational or even interventional cohort studies in this direction.

REFERENCES

[1] Fitzgerald KC,et al. Association of Vitamin D Levels With Multiple Sclerosis Activity and Progression in Patients Receiving Interferon Beta-1b. JAMA Neurol 2015

[2] Dörr J, et al. High-dose vitamin D supplementation in multiple sclerosis - results from the randomized EVIDIMS (efficacy of vitamin D supplementation in multiple sclerosis) trial. MultScler J - Exp Transl Clin 2020

Données génétiques	Génotype CC		Génotype CT		Génotype TT		Allèle C	Allèle T	
Données radiologiques	N=33	P	N=33	P	N=15	P	N=33	N=48	P
Lésions périventriculaires	29(88%)	0,35	32(97%)	0,13	13/15	0,47	29(88%)	45(94%)	0,35
Lésions sous-corticales	27(82%)	0,09	31(94%)	0,23	14/15	0,54	27(82%)	45(94%)	0,09
Lésions infratentorielles	23(67%)	0,91	23(67%)	0,91	11/15	0,78	23(67%)	34(71%)	0,91
Lésions des NGC	1(3%)	0,13	5(15%)	0,08	1/14	0,76	1(3%)	6(13%)	0,13
Lésions médullaires	23(67%)	0,77	19(58%)	0,09	13/15	0,08	23(67%)	32(67%)	0,77
Lésions confluentes	16(48%)	0,54	16(48%)	0,54	4/15	0,12	16(48%)	20(42%)	0,54
Prise de contraste	11(33%)	0,19	17(51%)	0,14	6/15	0,86	11(33%)	23(48%)	0,19
Black Hole	3(9%)	0,36	2(6%)	0,97	0/15	0,27	3(9%)	2(4%)	0,36
Atrophie cérébrale	8 (24%)	0,020	2(6%)	0,10	1/15	0,38	8(24%)	3(6%)	0,020
Atrophie médullaire	0(0%)	0,23	1(3%)	0,78	1/15	0,24	0(0%)	2(4%)	0,23

Table 1