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Molecular Study of Genomics Mutations in RNF213, ACTA2, GUCY1A3 Genes in Patients Moyamoya Syndrome in Human

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Abstract

In this study, we have analyzed 50 people. 20 patients Moyamoya disease and 30 Persons control group.The genes RNF213 on chromosome 17q25, ACTA2 on chromosome 10q23.3,GUCY1A3 on chromosome 4q32, analyzed in terms of genetic mutations made. In this study, people who have genetic mutations were targeted, with nervous disorders, Moyamoya disease. In fact, of all people with Moyamoya disease. 20 patients Moyamoya disease had a genetic mutation in the genes RNF213 on chromosome 17q25,ACTA2 on chromosome 10q23.3,GUCY1A3 on chromosome 4q32 Moyamoya disease. Any genetic mutations in the target genes control group, did not show.

Keywords: Genetic study; Moyamoya disease; Mutations in the genes RNF213,ACTA2, GUCY1A3

Introduction

Moyamoya disease is a disease in which certain arteries in the brain are constricted. Blood flow is blocked by the constriction, and also by blood clots (thrombosis) [1]. A collateral circulation develops around the blocked vessels to compensate for the blockage, but the collateral vessels are small, weak, and prone to bleeding, aneurysm and thrombosis. On conventional X-ray angiography, these collateral vessels have the appearance of a "puff of smoke" (described as "もやもや (moyamoya)" in Japanese) [1].

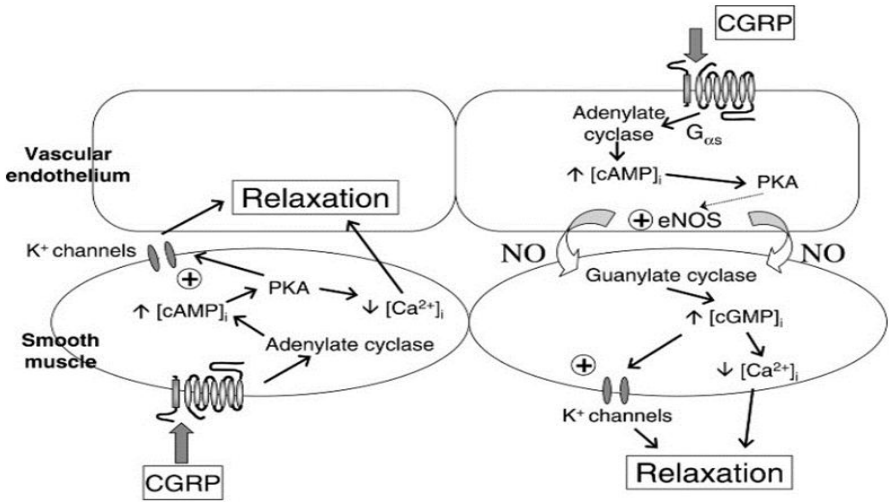


Figure 1: Schematic view of gene molecular the cycle of CGRP.

About 10% of cases of moyamoya disease are familial, and some cases result from specific genetic mutations. Susceptibility to moyamoya disease-2 (MYMY2; 607151) is caused by variation in the RNF213 gene (613768) on chromosome 17q25. Moyamoya disease-5 (MYMY5; 614042) is caused by mutation in the ACTA2 gene (102620) on chromosome 10q23.3; and moyamoya disease-6 with achalasia (MYMY6; 615750) is caused by mutation in the GUCY1A3 gene (139396) on chromosome 4q32. Loci for the disorder have been mapped to chromosome 3p (MYMY1) and chromosome 8q23 (MYMY3; 608796). See also MYMY4 (300845), an X-linked recessive syndromic disorder characterized by moyamoya disease, short stature, hypergonadotropic hypogonadism, and facial dysmorphism. and linked to q25.3, on chromosome 17". (Online Mendelian Inheritance in Man, omim.org/entry/252350) [2-7].

Materials and Methods

In this study, 20 patients with Moyamoya disease and 30 Personscontrol group were studied. Peripheral blood samples from patients and parents with written permission control was prepared. After separation of serum, using Real Time-PCR technique of tRNA molecules were collected. To isolate Neuroglial cells erythrocytes were precipitated from Hydroxyethyl Starch(HES) was used. At this stage, HES solutionin ratio of 1to 5 with the peripheral blood of patients and controls were mixed.After 60 minutes of incubation at room temperature, the supernatant was removed and centrifuged for 14 min at 400 Gera. The cells sediment with PBS (phosphate buffered saline), pipetazh and slowly soluble carbohydrate ratio of 1to2onficole (Ficol) was poured in the 480G was centrifuged for 34 minutes.Mono nuclear Neuroglial cells also are included,has a lower density than ficole and soon which they are based.The remaining erythrocytes has a molecular weight greater than fico land deposited in test tubes [8].

The supernatant, which contained the mono nuclear cells was removed, and the 400 Gera was centrifugedfor 12 minutes. Finally, the sediment cell, the antibody and Neuroglial cells was added after 34 minute's incubation at 5 °C, the cell mixture was passed from pillar LSMACS. Then the cells were washed with PBS and attached to the column LSMACSS pam Stem cell culturemedium containing the transcription genes RNF213,ACTA2,GUCY1A3,and were kept.

To determine the purity of Neuroglial cells are extracted, flow cytometry was used. For this purpose,approximately 80-110 × 103 Neuroglial cells were transfer red to1.5ml Eppendorf tube and then was centrifuged at 2000 rpm for 7minutes at time. Remove the supernatant culture medium and theremaining sediment, 100µl of PBS buffer was added. After adding 5-10µl PE monoclonal anti body to the cell suspension for 60 min at 4°C, incubated and read immediately by flow cytometry. For example,rather than control anti body Neuroglial cells PE,IgG1 negative control solution was used (Figure: 2-6) [9-10].

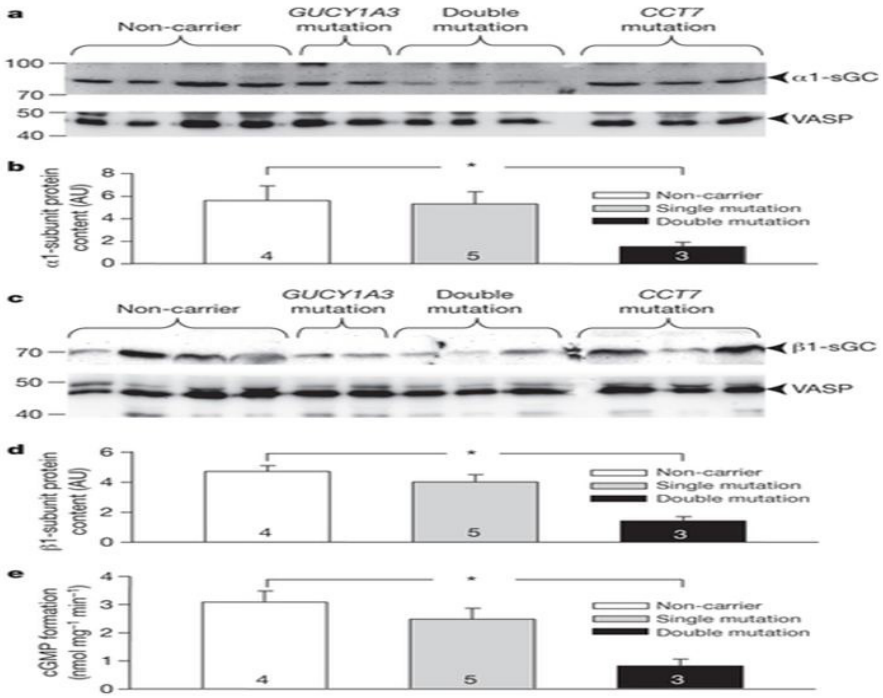


Figure 2: Schematic view of a column diagram and pattern band formed in GUCY1A3 gene in mutant and normal genes.

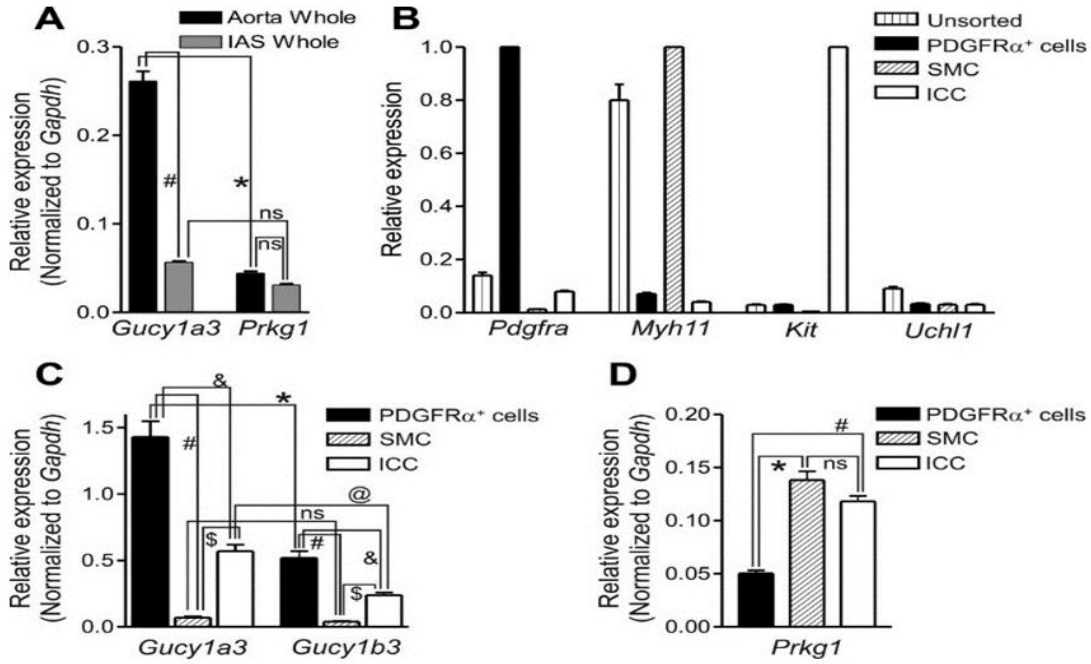


Figure 3: Schematic view of a column diagram of the expression of Genes GUCY1A3 and GUCY1B3 and PRKG1.

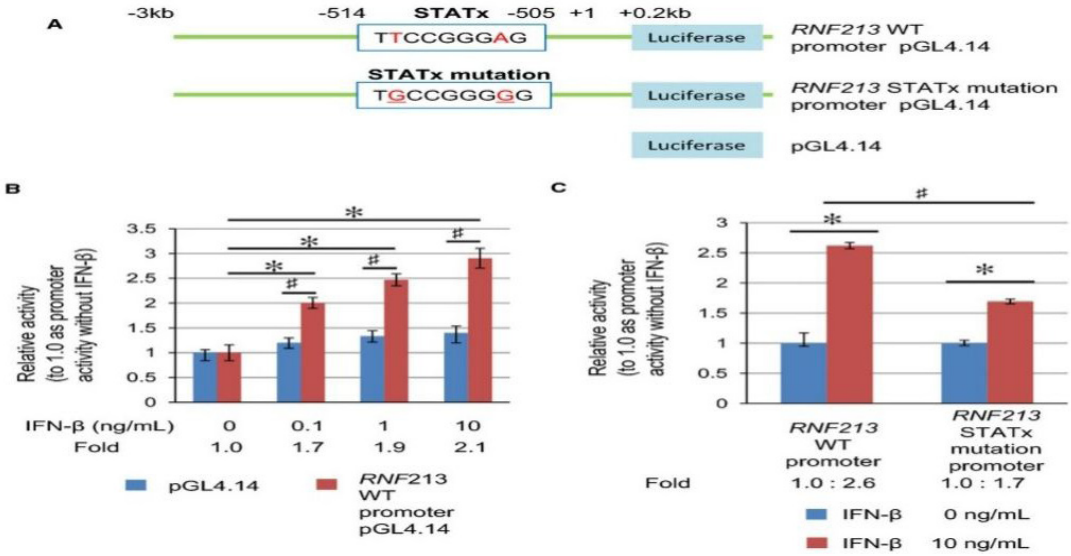


Figure 4: Schematic view of a column diagram RNF213 gene expression compared with normal and mutant genes.

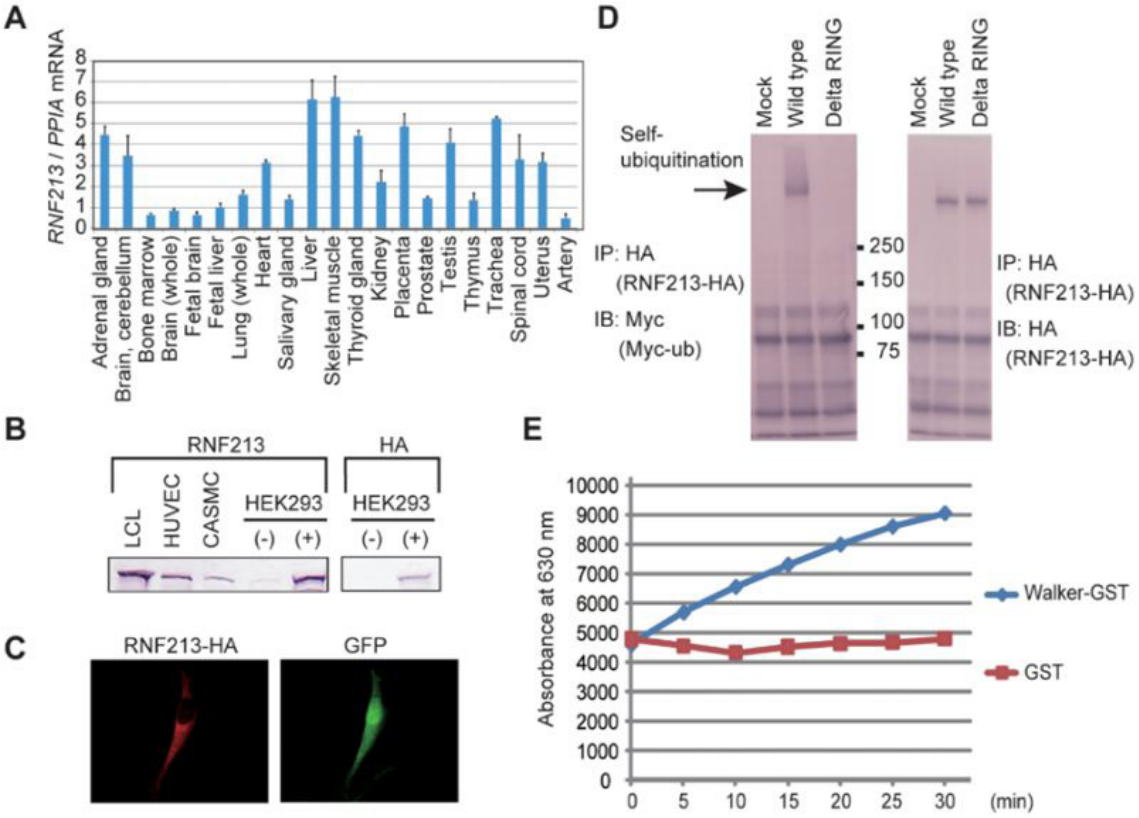
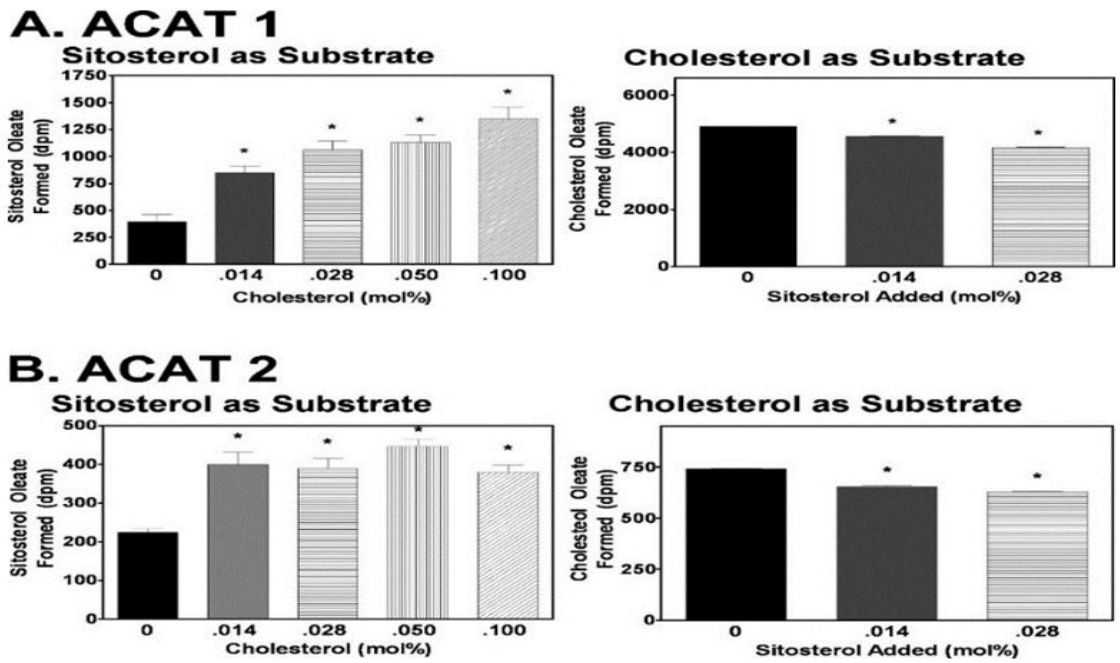


Figure 5: Schematic view of a column diagram and pattern band formed in RNF213 gene in mutant and normal genes.



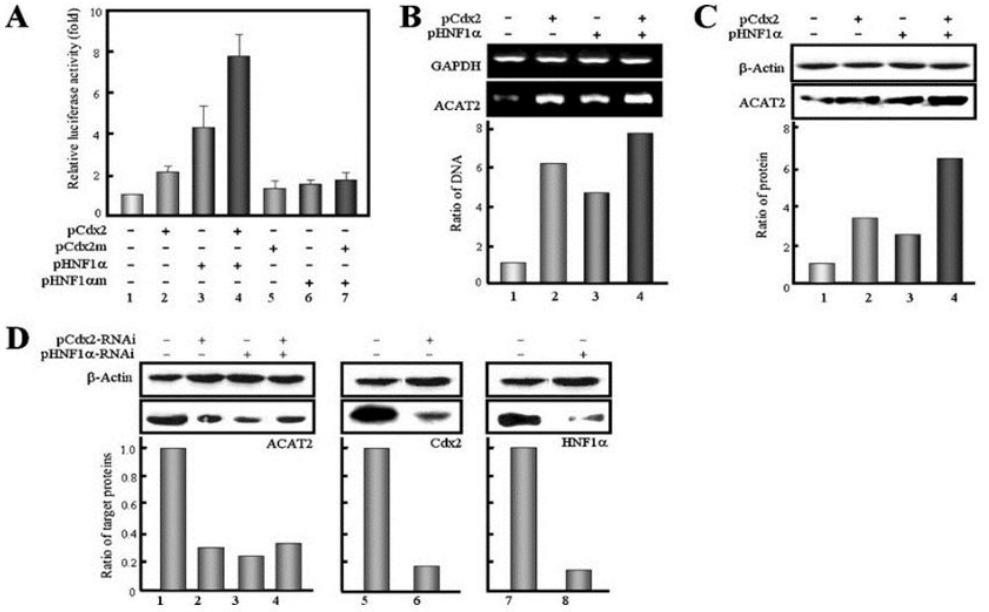


Figure 6: Schematic view of a column diagram and pattern band formed in ACAT2 gene in mutant and normal genes.

Discussion and Conclusion

According to the results of sequencing the genome of patients with Moyamoya disease, and the genetic mutations RNF213,ACTA2,GUCY1A3 genes found that about 96% of patients with Moyamoya disease, they have this genetic mutation. Patients with Moyamoya disease, unusual and frightening images in the process of Moyamoya disease, experience. Lot epigenetic factors involved in Moyamoya disease. But the most prominent factor to induce Moyamoya disease, mutations is RNF213,ACTA2,GUCY1A3 genes.These genes can be induced after birth and can also be induced in the adulthood. In this study, we could make several mutated genes that were involved in Moyamoya disease investigate. This study shows that nerve cells in humans can suffer different changes and yet these cells with genetic mutations. In the intervening neurological disease genes have a significant role. Despite being nothing more significant than the molecular changes in DNA for specific genetic diseases, but also epigenetic factors in the induction of these diseases are very important. We hope this study can be more research and more specific policies to improve the psychological state of patients with Moyamoya disease.

Acknowledgments

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