Assessment of Genetics Mutation in MECP2 Geneto Induce Rett Syndrome in Human of Tabriz, Iran

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Abstract

In this study, we have analyzed 20 people. 10 patients Rett disease and 10 persons control group. The gene MECP2, analyzed in terms of genetic mutations made. In this study, people who have genetic mutations were targeted, with nervous disorders, Rett disease. In fact, of all people with Rett disease, 10 patients Rett disease had genetic mutations in the genes MECP2 Rett disease. Any genetic mutations in the target genes control group did not show.

Key Words

Genetic study; Rett disease; Mutations the gene MECP2; Real Time PCR

Introduction

Rett Syndrome is a progressive neurodegenerative disorder that affects women almost exclusively, but in rare cases, men with Rett Syndrome have also been reported. The Rett syndrome is also known as a rare genetic disorder of the brain’s gray matter. Clinical features of the Rett Syndrome include small hands and feet, reduced head growth rate (microspheres), repetitive stereotypes of hand movements such as their own hands, due repeatedly inside the mouth. Infants with Rett Syndrome, usually 7 to 18 months after birth, reveal signs and symptoms of Rett Syndrome. People with Rett Syndrome are at risk of gastrointestinal disorders and experience up to 80% of seizure attacks. These people cannot learn any of the verbal skills, and usually 50% of these people do not have the ability to walk. Spinal curvature as a scoliosis and constipation disorder is very common and can be problematic [1-10].

Figure 1: Schematic representation of MECP2 gene in X chromosome

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Materials and Methods

In this study, 10 patients with Rett disease and 10 persons control 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 solution in ratio of 1 to 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 1 to 2 onficole (Ficol) was poured in the 480G was centrifuged for 34 minutes. Mono nuclear Neuroglial cells also are included, hasa lower density than ficole and soon which they are based. The remaining erythrocytes has a molecular weight greater than ficoleand deposited in test tubes.

The supernatant, which contained the mononuclear cells, was removed, and the 400 Gera was centrifuged for 12 minutes. Finally, the sediment cell, the antibody and Neuroglial cells was added after 34 minutes 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 culture medium containing the transcription gene MECP2, and were kept. Figure 3-8
Figure 3: Schematic View of the MECP2 mutation expression pattern versus healthy version.
**Figure 4:** Schematic representation of the band formation pattern in the mutated MECP2 gene compared to the healthy version.

(A) HONE-1, CPT30, KB, KB100

(B) MeCP2, P62

(C) TSA

(D) HONE-1, KB

(E) CPT30, KB100

(F) MeCP2

(G) MGMT

(H) β-Tubulin

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**Figure 5:** Schematic representation of the bond formation pattern in the mutated MECP2 mRNA compared to the healthy version.
Figure 6: Microscopic Image of the mutated MECP2 Gene Compared to the Healthy Version.
**Figure 7:** Microscopic Image of the mutated MECP2 gene in humans, mouse and rats.
Discussion and conclusion

According to the results of sequencing the genome of patients with Rett disease, and the genetic mutation MECP2 gene found that about 100% of patients with Rett disease, they have this genetic mutation. Patients with Rett disease, unusual and frightening images in the process of Rett disease, experience. Lot epigenetic factors involved in Rett disease. But the most prominent factor to induce Rett disease, mutation is MECP2 gene. This gene can induce the birth and can be induced in the adulthood.

References

1. Andrew S Davis (2010) Rett initially called this syndrome cerebroatrophic hyperammonemia, but the elevated ammonia levels in the bloodstream were later found to be only rarely associated with this condition.


