ISSN: 0976-3104
SUPPLEMENT ISSUE
Rahnavard et al.

THE IIOAB JOURNAL

ARTICLE OPEN ACCESS

# INVESTIGATING KINSHIP IN PRIMARY DYSTONIA PATIENTS WITHOUT MUTATION IN DYT1 GENE IN IRAN

**Sahereh Rahnavard, Mohammad Hamid*, Zahra Zand**

*Departmant of molecular medicine, Biotechnology Research Center, pastur institute of Iran, Tehran, IRAN*

## ABSTRACT

*Dystonia is one of the most painful diseases all over the world and it is debilitating too. Distribution of the disease is reported as 1 in 9000 of Ashkenazi Jews and 1in 16000 of non-Jews. There are some reports about the disease from different countries all over the world. Most of the reports are from pathological studies and genetic investigations in this disease have been limited to finding the limitation place.*

**Published on:** 25th– Sept-2016

KEY WORDS

*Dystonia, DYT1, General type, Multi focal type, writhing cramp, Focal type, Segmental type*

***Corresponding author: Email:** hamidi@pasteur.ac.ir **Tel.:**+989122858058

## INTRODUCTION

Dystonia is a disease which occurs due to pathological damage of muscles and leads to involuntary motions and paralysis [1]. A heterogeneous group of disorders with different reasons causes incidence of the disease. Dystonia is mostly accompanied by muscle contraction with repetitive spiral movements and abnormal contractions [2]. This disease is divided based on cause, beginning age and body distribution. It can be more divided to genetic and non-genetic groups based on the cause [3].

Dystonia is divided into several group according to the origin, onset age and body distribution [4].
On the basis of the origin, this disease is categorized into genetic and non-genetic groups [5].
The genetic group of Dystonia is also called Dystonia type I or first Dystonia [4,6]. The non-genetic group of Dystonia is also called Dystonia type II or second Dystonia [7].

Dystonia is classified according to onset age into early and late classes [8,9].The symptoms of early onset dystonia begin from childhood that usually presented as generalized, while that late onset dystonia occurs during adolescence in which head and neck usually involved(10).

Body distribution of dystonia can be exiseted as focal, local or generalized [11]. The symptoms of focal type presented in the 4-5th decade of the life and some of muscles were usually affected [12]. The generalized type of Dystonia started in the in the age less than 5 years and involved the most of muscles. It is the most inheritable type of Dystonia [13].

The average onset age is 12.5 year and almost occurs before the age of 28 in every affected individual. It appear as a focal dystonia and then distributed from hands or feet to over the body [14].

**Genealogy of the DYT1 patients**
**Among The patients without mutation in DYT1 gene in Iran**

28,98% of the patients (20 people) were children of family marriages.45% of them (9 people) had parents with third degree kinship and 105 of them had parents with far kinship. Age range of them had ben from birth to 48 years old. 45% of the patients (9 people) had general distribution type, 255 ( 5 people) had multi focal type, 5% ( 1 people) had writhing cramp, 5% (one person) had focal type and 20% ( 4 people) had segmental type. Patients

with hemi-dystonia type didn't have family relationship. Beginning of the disease in most of the patients had been from right side. In 45% of them it has begun form hand, 25% from leg (5 people), 20% (4 people) from face. The diagram of body distribution in patients with family relationship has drown in **[Figure- 1].**

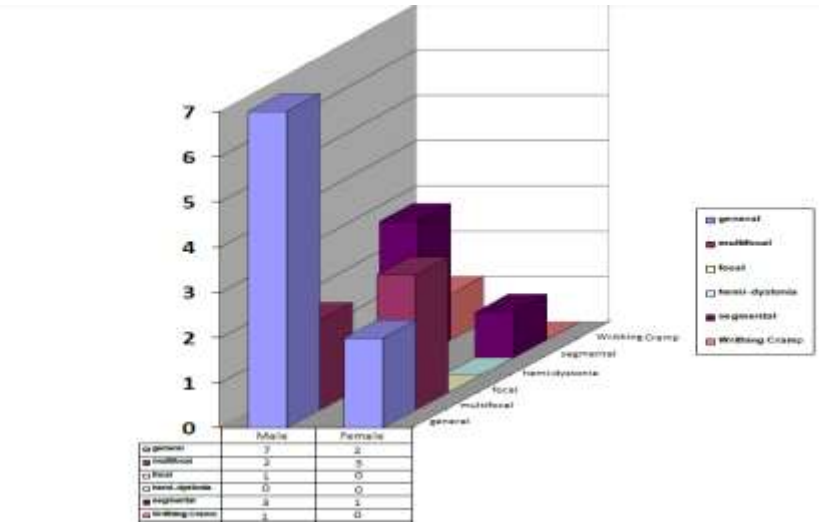


**Fig: 1. Body distribution in patients with family relationship**

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## Kinship in General type patients

From among general type patients, nine of them had family relationship which six of them are male and three are female. Two male and a female had third degree kinship and others have far relationship . From among these patients three of them had beginning age of zero to four years old and three of them had beginning age of four to eight years old, three of them were over eight at the beginning of the disease **[Figure-2]**.

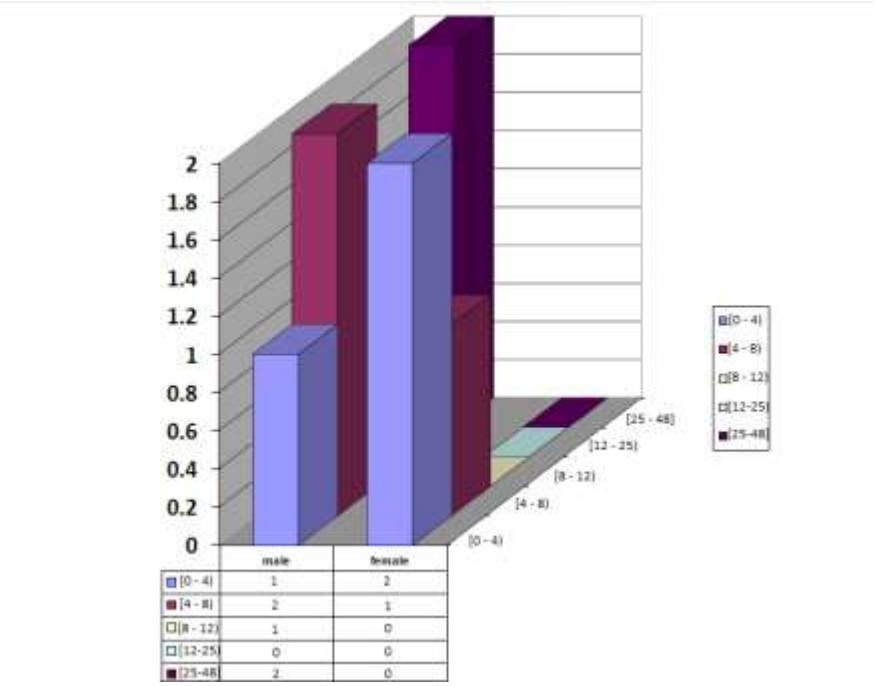


**Fig:2. Family relationship in general type patients**

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ISSN: 0976-3104



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**Patients of Dystonia Family relationship in Multi focal patients**

From among multi focal patients five of them had family relationship. Two of them are male and three of them are female. One male and two female had third degree family relationship and others have far relationship . Among these patients no beginning age from zero to four years old has been seen. One of them had the beginning age of four to eight and four of them had beginning age of over eight years old **[Figure-3]**.

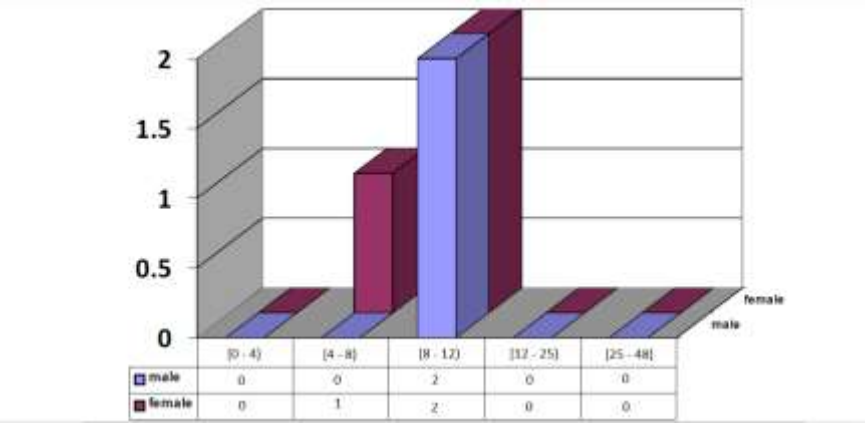


**Fig: 3. Family relationship in multi-focal patients**

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**Family relationship in multi focal patients**

From among focal type one person (one male) had family relationship with third degree relationship and incidence age is high .

**Family relationship in segmental patients**

From among segmental type patients four of them had family relationship. Three of them are male and one of them is female. Family relationships were third degree relationship and all had far family relationship. From among the patients one of them had the beginning age of zero to four years old and three of them had over eight years old age **[Figure-4]**.

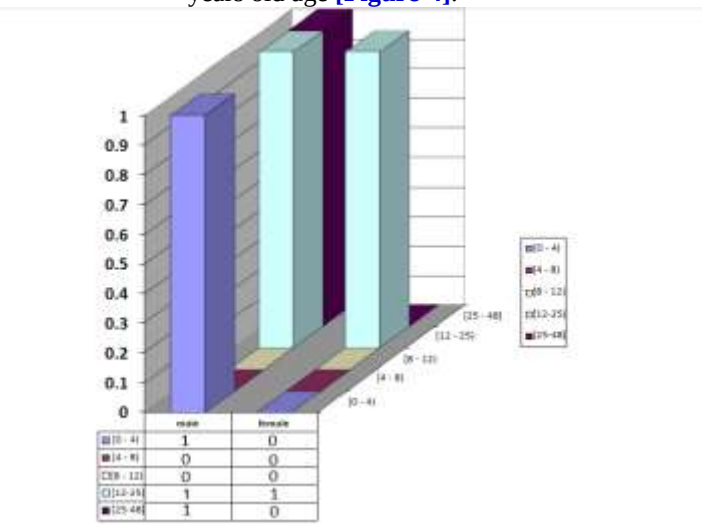


**Fig: 4. Investigating family relationship in segmental type patients**

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THE IIOAB JOURNAL
ISSN: 0976-3104



**Family relationship in writhing cramp patients**

Only writhing cramp patients show third degree family relationship .

## CONFLICT OF INTEREST

The author declares having no competing interests.

## ACKNOWLEDGEMENT

None

## FINANCIAL DISCLOSURE

None

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