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RESEARCH ARTICLE

Evaluating Age and Gender of Iranian Dystonia Patients with no Mutation in DYT1 Gene

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Abstract

Dystonia is a neuronal- motional disease that occurs due to pathological injuries to muscle and leads to involuntary and tick-like movements because of long-term muscle contraction and finally leads to paralysis and unusual situations in body that can occur in one or a group of muscles.

Introduction

This disease is almost known with muscle contraction and repetitive twisted movements that involves agonist and antagonist muscles. It has been observed firstly in Ashkenazi Jews population with prevalence of 1 in 9000 people while in other population prevalence of 1 in 16000 with low fitness around 30% has been reported.

Environmental factors causing this disease are not yet known in some cases. This type is called pr Dystonia type I. Environmental factors for Dystonia type II are known including spinal and brain damages, injuries, insufficient oxygen at birth, infections, drugs side-effects, increased bilirubin at childhood and some local diseases inside skull. Dystonia is included in early and late classes based on onset age. Body distribution can be focal, local, generalised, multifocal, hemidystonia and segmental.

Dystonia I

Some patients with Dystonia I have mutation in certain genes such as DYT1 and DYT6. These genes are inherited as autosomal dominant. The other genes have shown different inheritance. The most painful type is the early onset Dystonia I that involves all muscles and mostly shows generalized type. DYT1 and DYT6 are mainly responsible for this disease.

Here in this study we have evaluated the age and gender of 69 Iranian Dystonia type I patients without any mutation.

Average onset age for DYT1

Average onset age for these patients was 13.4 years old, approximately two times more than DYT1+.

Onset age before age 12

In studied patients, 31.88% (22 people) have shown the disease before age 12, of which 10.14% (7people) were before age 4, 13.04 % (9 people) were between ages 4 to 8 and 7.24 % (5 people) were before age 12. Men to women ratio for patients in this age range were 1:0.8. Also, number of affected men in age interval 7-8 was three times more than women.

Affected women before age 12

38.42 % (10 people) of affected women have shown the disease before age 12, of which 70 % (7 people) were before age 8 and 30 % (3 people) were between ages 8 to 12.

Affected men before 12

30.23 % (13 people) of affected men have shown the disease before age 12, of which 69.23 % (9 people) were before age 8 and 30.76 % (4 people) were between ages 8 to12.

Onset age more than 12 years

28.98 % (20 people) of patients have shown 12-48 years old onset age, of which 45 % (9 people) were women and 55 % (11 people) were men. The disease in lower ages was shown by women more than men, but at older people number of affected men were more than women. The onset age has not been identified for 41.16 % of patients.

Body distribution

Generalized

Generalized distribution was shown in 26.08 % (18 people) of patients, of which 61.11% (11 people) and 38.88% (7people) were men and women, respectively and men to women ratio was 1:0.8. Different age range has been observed in this type of body distribution. However, women's onset age was mostly at childhood and only one case has shown the disease at age 48, while men`s onset age has shown a normal distribution.

Focal

10.14 % (7 people) have shown focal type, of which 57 % (4 people) and 42 % (3 people) were men and women respectively and men to women ratio was 1:0.75. The onset age for this type was 21.5± 6.5 and as shown it can be seen mostly at adulthood without any early type.

Multi focal

Multi-focal distribution was the most frequent form after generalized type. Multi focal type was seen in 14.70 % (9 people) of patients, of which 66.66% (6 people) and 33.33 % (3 people) were men and women respectively with men to women ratio 1:0.5. The onset age was 14±10 years which only one patient has shown the disease as early type at age 4.

Segmental

10.14 % (7 people) have shown segmental distribution that 42.85 % (3 people) and 57.14 % (4 people) of them were men and women respectively and men to women ratio was 1:1.35. This is the only type that is more frequent in women than men and is shown just at adulthood. Only one early type patient has a segmental distribution.

Hemidystonia

5.7 % (4 people) of patients have shown Hemidystonia distribution, of which 75% (3 people) and 25 % (1 people) were women and men respectively with men to women ration 1:3.

Writhing cramp

This type is only seen in one man with onset age 43years.

Discussion and conclusion

In this study, men to women ratio in generalized and multi focal were 1:0.8, while men to women ratio have decreased in other types. The onset age at childhood can be seen mostly in multi focal patients.

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