HEREDITARY CONGENITAL FACIAL PALSY

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ABSTRACT

Hereditary congenital facial palsy (HCFP) is a rare disorder. We report the case of 20 year old woman who presented with right sided facial weakness since her infancy. Among five generations of her family, 12 other members had facial palsy. Her detailed clinical assessment revealed no abnormality other than right sided facial paresis. A diagnosis of autosomal dominant hereditary congenital facial palsy was made.

Keywords: Autosomal dominant, Facial palsy, Five generations.

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INTRODUCTION

Hereditary congenital facial paresis (HCFP) is an isolated dysfunction of facial nerve. It belongs to a group of congenital diseases known as congenital cranial dysinnervation disorders (CCDDs), characterized by abnormal eye and facial movements[1].

The disorders belonging to CCDDs include Duane syndrome, Möbius syndrome, horizontal gaze palsy, congenital ptosis and congenital facial palsy. Although Möbius syndrome and HCFP share some clinical features, they are different entities.

Möbius syndrome is a developmental disorder of lower brainstem associated with facial palsy and impairment of ocular abduction. Hereditary congenital facial weakness results from maldevelopment of facial nucleus and its' nerve. There may be complete or partial absence of facial nerve on one side or both sides[2].

CASE REPORT

A 20 year old female patient with congenital right sided facial weakness was referred to Neurology Clinic of Military Hospital (MH) Rawalpindi by Medical Officer of Heavy Industries Taxila (HIT) in Dec 2012. She produced a hand written list of their family members (made by her father, an employ of HIT) among 5 generations, suffering from congenital facial weakness (fig-1). According to this list 12 other

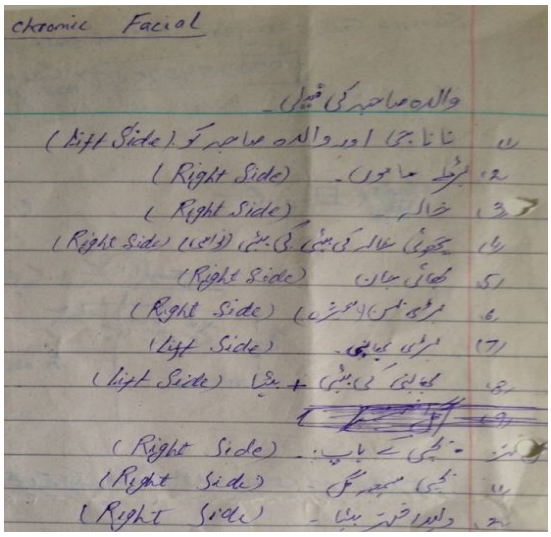

Figure-1: List of the family members having facial weakness.

members of her family suffered from congenital facial weakness:

1. Maternal grandfather of her father (Left) (1st Generation).
2. Mother of her father (Left) (2nd Generation).
3. One maternal Uncle of her father (Right) (2nd Generation).

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4. One Maternal aunt of her father (Right) (2nd Generation).
5. Her Father (Right (3rd Generation).
6. Her paternal uncle (Right) (3rd Generation).
7. Her paternal aunt (Right) (3rd Generation).
8. Her brother (Right) (4th Generation).
9. Daughter of maternal cousin of her father (Right) (4th Generation).
10. Elder niece of her father (left) (4th Generation).
11. Daughter of niece of her father (left) (5th Generation).
12. Son of niece of her father (Left) (5th Generation).

Neurological examination revealed unilateral lower motor neuron paresis of right facial nerve. Detailed clinical assessment revealed no other abnormality. A diagnosis of autosomal dominant hereditary congenital facial palsy was made. She was explained the prognosis of the disease. Since then she was lost for follow up.

DISCUSSION

HCFP is a rare autosomal dominant inherited disorder affecting some families. Two large HCFP families i.e. HCFP1 and HCFP2 were first described in two Dutch families. Linkage analysis in these two families identified two different loci, 3q21–22 in HCFP1 and 10q21–22 in HCFP2, indicating genetic heterogeneity for this disorder.

The phenotype for HCFP1 family is an asymmetric, mostly bilateral, weakness of facial muscles with a penetrance of 95%. The phenotype of HCFP2 family is often characterized by an asymmetrical, unilateral or bilateral facial weakness with a penetrance of 60%. In addition to facial weakness congenital deafness may also be present[3].

Michielse et al describe a third large autosomal dominant HCFP family originating from Pakistan. Linkage analysis identified the locus at 3q21–22, like the Dutch HCFP1 family[4].

In a large Dutch family in which 46 persons in 6 generations had congenital facial paralysis Kremer et al examined 31 family members, including 20 affected persons. The proband had asymmetric facial weakness. He was born with facial weakness similar to his grandmother and many of his siblings. His obligate carrier mother had no evidence of facial muscle weakness[5].

In HCFP an appropriate history and physical examination is sufficient to make a diagnosis. An extended physical examination is needed to exclude other congenital malformations. Imaging and neuromuscular testing may be necessary for treatment planning.

CONFLICT OF INTEREST

This study has no conflict of interest to declare by any author.

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