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Research Article

Clinical Features of Congenital Anosmia

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Abstract: The objective of the study was to evaluate the clinical features of congenital anosmia. We retrospectively analyzed 205 patients at our hospital for olfactory disturbances over a 4-year period. The overall prevalence of congenital anosmia was found to be 3.4% (7/205). Magnetic resonance imaging of the brain revealed the absence of the olfactory bulbs, tracts, and hypoplasia of the olfactory sulci in all patients. Two patients who had hypogonadotropic hypogonadism were diagnosed with Kallmann syndrome of the patients who complained of olfactory disturbances, the prevalence of congenital anosmia was found to be 3.4%. These results may aid in explaining clinically rare conditions. We emphasize that young patients should be carefully observed for the development of secondary sex characteristics, and hormone replacement therapy should be considered in patients of a fertile age.
Keywords: Olfactory disturbance, Congenital anosmia, Kallmann syndrome, Prevalence, Magnetic resonance imaging, Hypogonadotropic hypogonadism

INTRODUCTION

The most common causes of olfactory dysfunction include allergic rhinitis, chronic rhinosinusitis, and upper respiratory infections. Other potential causes include head trauma, neurodegenerative diseases, medications, and congenital anosmia.

Congenital loss of smell, i.e., the inability to recognize odors since birth, can be classified into two categories: Group 1 comprises about 12% of patients, which commonly exhibit familial loss of smell associated with major congenital abnormalities, including hypogonadotropic hypogonadism as well as genetic, somatic, and neurologic abnormalities [2]; Group 2 comprises the majority (88%) of patients with a congenital loss of smell [2]. Group 2 patients show the same degree of loss of smell as Group 1 patients; however, they generally lack a familial association, have normal gonadal function, and do not exhibit other somatic abnormalities [2]. We have previously reported on patients with isolated congenital anosmia [3] and Kallmann syndrome [4].

Recently, several reports have been published concerning the possibility of diagnosing morphological abnormalities of the olfactory bulbs, tracts, and sulci by using magnetic resonance imaging (MRI) [2-15].

In this study, we report the clinical features and MRI findings of patients with congenital anosmia.

MATERIALS AND METHODS

We conducted a retrospective survey of patients who visited the Jichi Medical University hospital for olfactory disturbances from April 2010 through to March 2014. A total of 205 patients were analyzed. An otolaryngological endoscopic examination and T&T olfactogram were performed. In addition, MRI of the brain was evaluated.

RESULTS

The overall prevalence of congenital anosmia was 3.4% (7/205). All cases are listed in Table 1. The age of diagnosis varied from 6 to 45 years in our study. Out of seven patients with congenital anosmia, four were men and three were women. MRI detected the absence of the olfactory bulbs, tracts, and hypoplasia of the olfactory sulci in all patients. Of the seven patients, three patients showed hypogonadotropic hypogonadism. Based on further urological and endocrinological consultations, two patients (Cases 3 and 4) were diagnosed with Kallmann syndrome. These patients were put on a treatment of intramuscular injections of testosterone enanthate. One patient (Case 1) developed secondary sex characteristics during the observation period.

Table 1: Cases of congenital anosmia

Case	Age (years)	Sex (M/F)	T&T Olfactogram	Olfactory bulbs	Olfactory sulci	Hypo-gonadism	Kallmann syndrome
1	6	F	scale out	absent	hypoplastic	+	−
2	15	F	scale out	absent	hypoplastic	−	−
3	18	M	scale out	absent	hypoplastic	+	+
4	28	M	scale out	absent	hypoplastic	+	+
5	9	F	scale out	absent	hypoplastic	−	−
6	23	M	scale out	absent	hypoplastic	−	−
7	45	M	scale out	absent	hypoplastic	−	−

DISCUSSION

Congenital anosmia is extremely rare. Hashimoto *et al.* [16] identified three patients (0.52%) with congenital anosmia among 578 patients with olfactory disturbances. In the present study, the overall prevalence of congenital anosmia was 3.4%.

Kallmann syndrome is a rare genetic disorder with an estimated prevalence of 1 in 10,000 males and 1 in 50,000 females [10]. Yousem *et al.* [5] reported eight patients with Kallmann syndrome among 25 patients with congenital anosmia. Aiba *et al.* [6] reported two patients with Kallmann syndrome among nine patients with congenital anosmia. In the present study, two patients had Kallmann syndrome among seven patients with congenital anosmia. In addition, Jagtap *et al.* [11] evaluated 41 patients with hypogonadotropic hypogonadism. According to their report, 25 patients had Kallmann syndrome and 16 were normosmic.

CONCLUSION

We propose that conventional MRI may alert the clinician to the possibility of a congenital olfactory dysfunction. We recommend that early consultation with a pediatrician or an endocrinologist for appropriate support and reassurance should be offered to those who have hypogonadotropic hypogonadism. Finally, we emphasize that young patients should be carefully observed for the development of secondary sex characteristics, and hormone replacement therapy should be considered in patients of a fertile age.

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