

A Case of Kallmann's Syndrome with Hypoplasia of Olfactory Bulb

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= Abstract =

Kallmann's syndrome has both a general and specific connotation in describing general condition of gonadotropin – releasing hormone(GnRH) deficiency or a particular cluster of anomalies associated with primary eunuchoidism. The familial occurrence of hypogonadotropic hypogonadism associated with anosmia, color blindness, synkinesia, and mental defect is the classic Kallmann's syndrome. Interestingly, anosmia, or lack of smell, was not found in the absence of gonadal deficiency in the original study of this disorder. This disorder was found on both sexes, but the male to female ratio was 11 : 1, and Kallmann's syndrome is more often listed under disorders of male hypogonadism for this reason.

Gross anatomy has shown disorders of the olfactory bulbs associated with Kallmann's syndrome and it was demonstrated a failure of GnRH – containing cells to migrate from the olfactory placode to the hypothalamus and preoptic area.

We have experienced a case of Kallmann's syndrome which

showed a hypoplasia of olfactory bulb in MRI during the workup of primary amenorrheic patient. So we report this case with a brief review of literatures.

Key Words : Kallmann's syndrome, Olfactory bulb

1,000 1 .
(epiphysis) (trunk)
가 가 .
(cryptorchidism)
testosterone, estradiol
가 , , X
가 , X
22.3 .
가
(isolated gonadotropin deficiency)
가
16
18 .

25

가

2 3 3

2

2

37°C, 80 / 159cm, 52kg, 130 / 70 mmHg, Tanner 1

estradiol : < 13 pg/ml, FSH : < 1.0 mIU/ml, LH : < 1.0 mIU/ml, DHEA - S : 278.9 µg/dl, 17 - OHP : 1.89 ng/ml, testosterone : 1.3 ng/ml, free testosterone : 0.7 pg/ml . TSH : 3.50 µU/ml, prolactin : 13.9 ng/ml

(olfactory sulcus)

(olfactory bulb)

(Fig. 1). KALIG -

1 (fluorescent in situ hybridization : FISH) 46,XX

가

(hypogonadism)

(anosmia)

1944

가

(sporadic)

가

X

(heterogeneity)

(White et al.,

1983 ; Hermanussen and Sippell, 1985).

1 / 10,000,

1 / 50,000

(Jones and Kemman,

1976). 가

5 - 6

X

가 가

(isolated gonadotropin deficiency) 가
(Naftolin et al., 1971).

(Santen, 1991).

(fertile eunuch syndrome)

(gynecomastia), (micropenis),
(cryptorchidism) (Turner et al., 1974).

(analogue)
가 (Burris et al., 1988 ; Griffin and Wilson, 1992).

(anosmia)
(hyposmia) (olfactory bulb)
(olfactory tract)
(Knorr et al., 1993 ; Truwit et al., 1993 ; Yousem et al., 1993).

가 (olfactory gyrus)
(olfactory bulb) (olfactory ventricle)
(olfactory sulcus)
(forebrain)

가
(synkinesis), (sensorineural deafness),
(cerebellar ataxia), (Wegenke et al.,
(spastic paraplegia), 1975 ; Tuck et al., 1983 ; Kertzman et al., 1990).
(pes cavus),
(Sunohara et al., 1986 ; Schwankhaus et al., 1989).

(neuronal migration) 가

가 (Schwanzel – Fukuda and Pfaff, 1989 ; Wray et al., 1989).

(olfactory placode) (ectoderm) (cribriform plate) (meningeal tissue) (mitral cell) 가 (dendrite) (synapsis)

(axon) (isolated hypogonadotropic hypogonadism) -

100 µg
500 µg

가 (pulsatile) 가 (Naftolin et al., 1971).

가

(body mass index)가

(Albanese and Stanhope, 1995 ; Finkelstein et al., 1996).

X

가

KAL 가

가

가

가

X

가

가

가

(incomplete penetrance)

가

X

KAL

가

KALP

KALP

가

KALP

가

testosterone

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