

# KALLMANN SYNDROME CAUSED BY SOX2 GENE MUTATION

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## INTRODUCTION

SOX2 gene encodes SOX2 transcription factor who maintains the pluripotency of stem cells, mediates the differentiation of neurons, and plays an important role in the development of the eye, forebrain, hypothalamus and pituitary. It is known to cause **idiopathic hypogonadotropin hypogonadism** but has never been found to be accompanied by an absence of the olfactory bulb. Now we present **a case of IHH with absence of olfactory bulb caused by SOX2 gene missense mutation.**

## AIM

To enhance the understanding of the clinical phenotypes of patients with SOX2 gene mutations and **expand the pathogenic gene pool of Kallmann syndrome.**

## METHOD

A retrospectively analysis of the clinical manifestations and the laboratory results of a child with Kallmann syndrome caused by SOX2 gene mutation confirmed by **whole exome-gene sequencing.**

## RESULTS

LHRH stimulation test	0'	30'	60'	90'	120'
LH(mIU/ml)	0.06	0.59	0.5	0.41	0.38
FSH(mIU/ml)	0.43	2.32	2.47	2.9	2.94

Table 1. LHRH stimulation test result showed hypogonadotropin hypogonadism.

basal test after 4 months pulsatile GnRH treatment	T(ng/ml)	DHT(pg/ml)	FSH(mIU/ml)	LH(mIU/ml)
before treatment	<0.13	NA	5.47	1.97
after treatment	0.16	255.60	0.43	0.06

Table 2. Sex hormone basal level before and after 4-month pulsatile GnRH treatment.

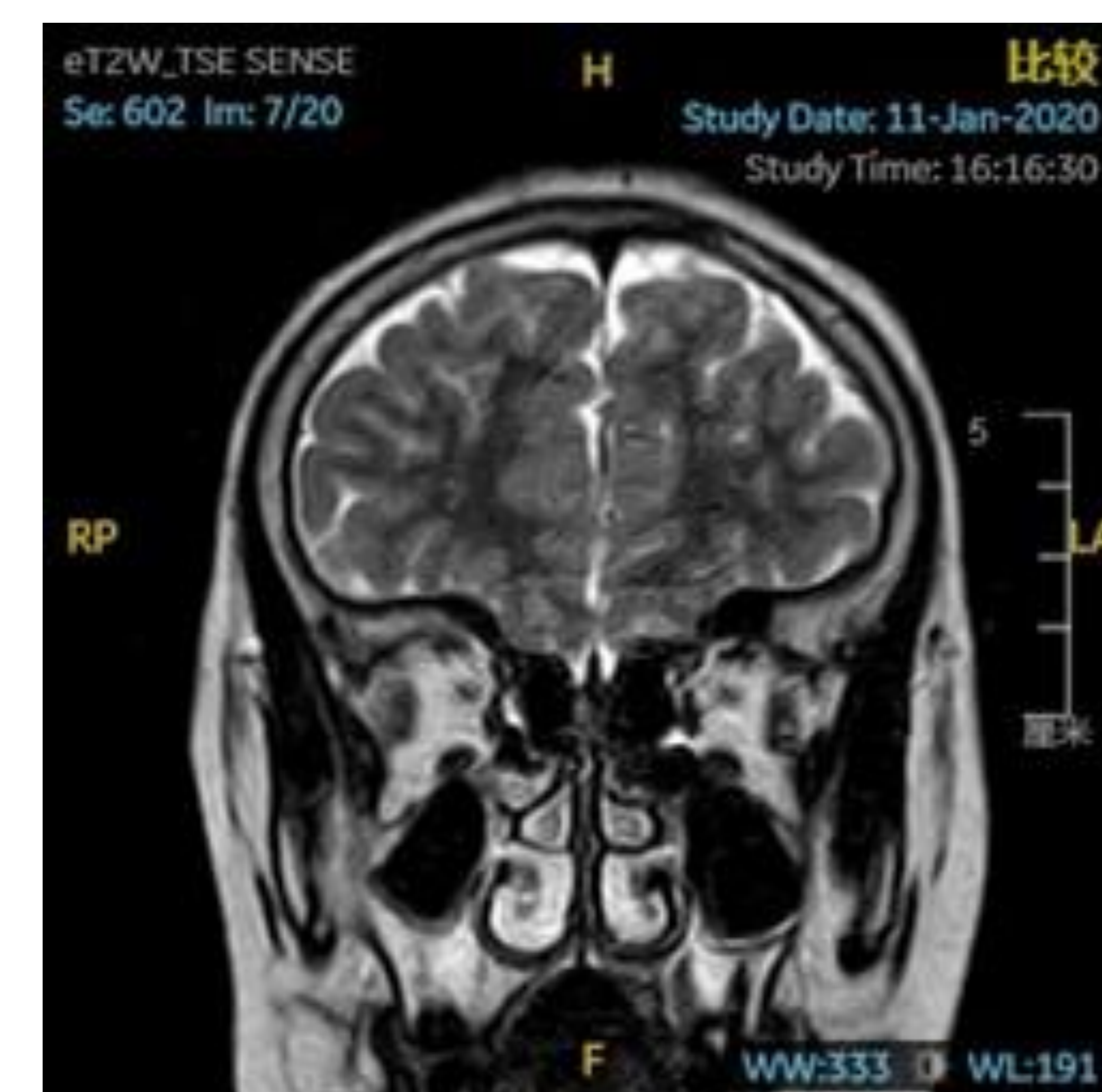


Fig 1. The MRI image of the patient showing absence of olfactory bulb.

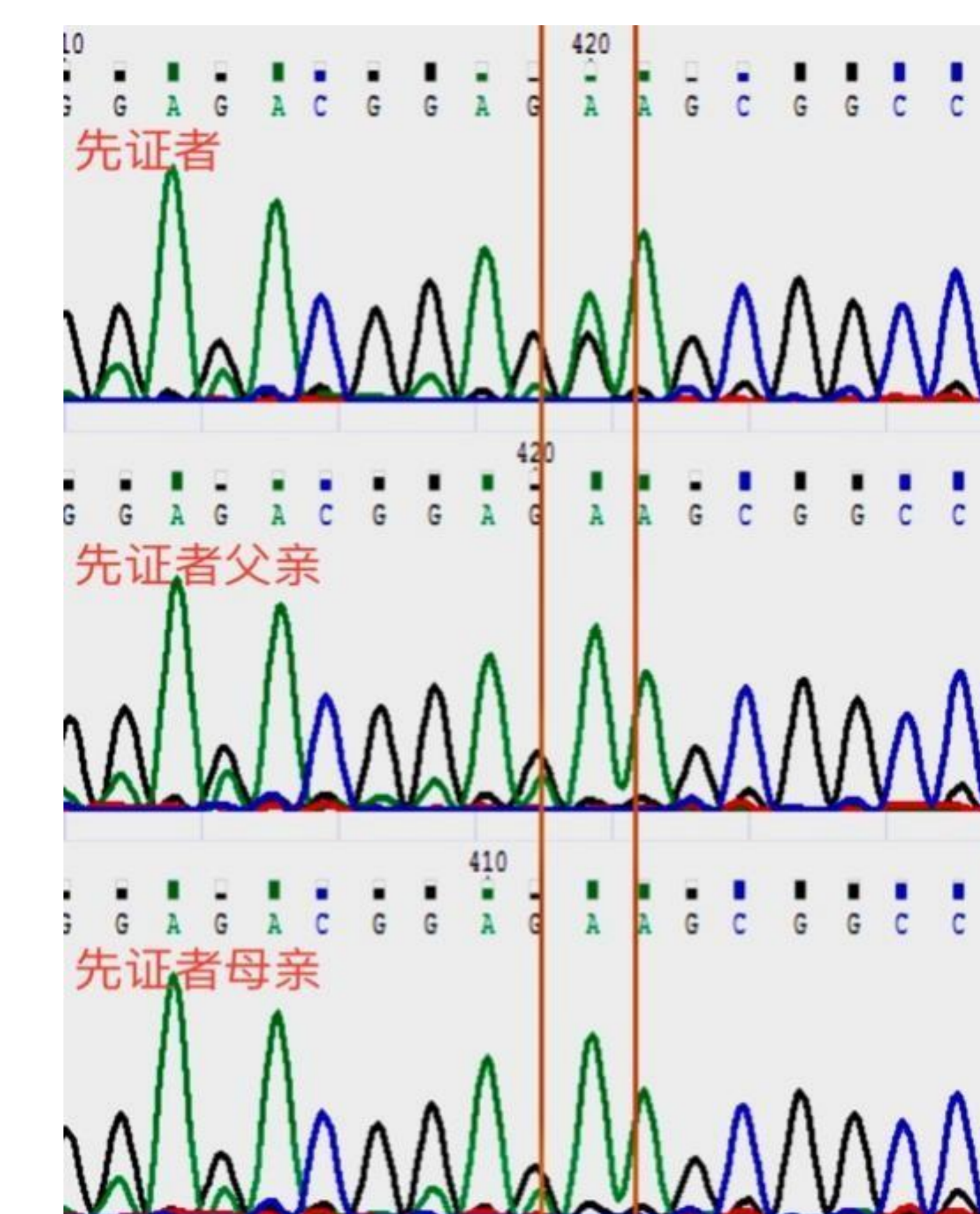


Fig 2. Sanger sequencing result of the patient and his parents after whole exome-gene sequencing showing SOX2 gene mutation

## CONCLUSIONS

- The SOX2 gene is involved in the development of the eyeball, the olfactory bulb and gonads. Mutations in this gene can lead to anosmia and hypogonadism.
- The mutation site of this case has not yet been reported, and this is the first reported case of Kallmann syndrome caused by SOX2 gene mutation, expanding the clinical phenotype of SOX2 gene mutations and the pathogenic gene pool of Kallmann syndrome.
- The whole exome-gene sequencing methods is helpful in the molecular diagnosis of such genetic heterogeneous diseases.

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