

Supplementary Data.

Methods

Pituitary magnetic resonance imaging (MRI): MRI scans were performed in a 1.5 Tesla unit Signa GE, Milwaukee, Wisconsin using T1 weighted sagittal and coronal scans with TR:350 ms and TE:20 ms. The coronal images were obtained using 3.0 mm thickness slices with 10% gap before and after administration of 0.1 mMol/L of a paramagnetic contrast (gadolinium). Patients with nonvisible pituitary stalk associated to ectopic posterior pituitary, optic dysplasia, other CNS malformation or surgical interventions were not included.

Supplemental Table 1: Haplotyping analysis of Brazilian and Japanese patients harboring p.E382E *GHRHR* mutation. Results from (a) seven microsatellites flanking the *GHRHR* and (b) five *GHRHR* SNPs including the possible p.E382E (c.1146G>A) mutation site.

	Patient 3		Father of Pt 3		Mother of Pt 3		Japanese Pt 1		Japanese Pt 2	
D7S516	313.52	317.47	313.50	317.46	317.45	317.45	317.52	317.52	317.42	317.42
D7S2496	232.43	224.90	232.37	224.80	224.89	226.78	213.62	213.62	213.66	213.66
<i>GHRHR</i>										
D7S2252	255.87	255.87	256.05	254.14	255.94	259.88	254.10	254.10	254.09	254.09
D7S484	107.07	107.07	107.07	107.07	107.05	107.05	107.08	107.08	107.12	107.12
D7S510	84.98	89.02	84.98	87.00	88.97	93.04	89.02	89.02	89.07	87.05
D7S691	150.53	144.02	150.43	146.20	143.96	141.82	148.43	148.43	148.33	150.53
D7S2427	248.69	223.90	248.61	223.80	223.87	229.60	223.89	223.89	223.93	223.93

(a)

	Patient 3		Father of Pt 3		Mother of Pt 3		Japanese Pt 1		Japanese Pt 2	
<i>GHRHR</i> 5'										
-261C>T	T	C	T	C	C	C	C	C	C	C
-235C>T	T	C	T	C	C	C	C	C	C	C
-45C>T	C	C	C	C	C	C	C	C	C	C
c.975-26C>T	G	G	G	G	G	G	G	G	G	G
c.1146G>A (p.E382)	A	G	A	G	G	G	A	A	A	A

GHRHR 3'

(b)

Microsatellites/*GHRHR*-R/position:

D7S516 (313-317)/28197695 bp, D7S2496 (213-232)/29566467bp, *GHRHR*/3100363 bp; D7S2252 (254-259)/32073581 bp, D7S484 (107)/35284906 bp; D7S510 (84-89)/39189712; D7S691(143-150)/42029531; D7S2427 (223-248)/44964494.

GHRHR SNPs:

-261C>T (rs2302019), c.-235C>T (rs230220), c.-45C>T (rs2302021), c.975-26G>A (rs4988504).