

Table 2 Summary of clinical findings in Kallmann syndrome patients

Novel *KALI* mutation p.Trp204*

| | Patients | | Novel <i>KALI</i> mutation p.Trp204* | |
|---|---|------------------------------------|--------------------------------------|----------------------|
| | K.01 | K.02 | K.03 | K.04 |
| Sex | Male | Male | Male | Male |
| Age (years) | 30 | 27 | 26 | 23 |
| Sense of smell | Hyposmia | Anosmia | Anosmia | Anosmia |
| Sexual maturation* (Tanner stage) | G4P4B3 | G4P4B3 | G4P4B3 | G2P2B3 |
| Testicular volume and cryptorchidism** right/left | C-R/3 mL | 4 mL/C-L | C-R/4 mL | C-R/C-L |
| Renal malformation | Right kidney agenesis | Left kidney agenesis | Right kidney agenesis | Left kidney agenesis |
| Dental agenesis | First, second and third lower right molars and first lower left molar | First and third lower right molars | Third lower right and left molars | ND |
| Synkinesis | Bimanual | Bipodal | Bimanual | ND |
| Short fourth metacarpal | Right/left | No | No | ND |

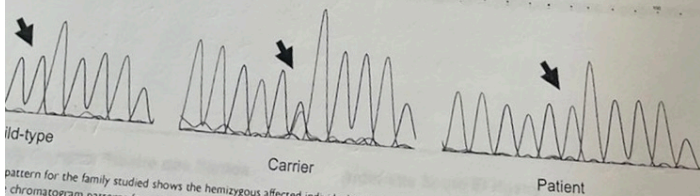
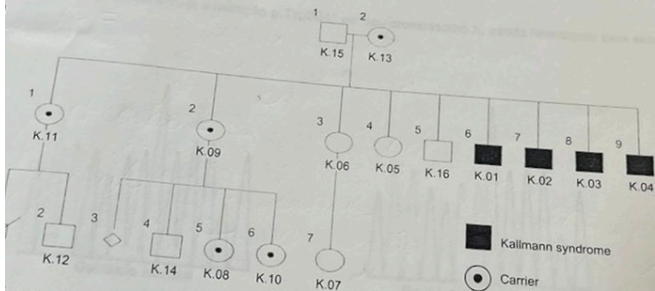
Notes: *G, external genitalia; P, pubic hair; B, breast development; **C-R, right cryptorchidism; C-L, left cryptorchidism.

Abbreviation: ND, no data available.

renal agenesis, cryptorchidism, synkinesis and dental agenesis. Nystagmus, ptosis, auricular dysgenesis and cleft lip/palate were not found in any patient.

The molecular analysis detected the transversion G to A at position 612 on exon 5 of the *KALI* gene.

This mutation was hemizygous in the four affected brothers. Five females heterozygous for this mutation were identified in the family, including the patients' mother (Figure 1). The same mutation was not found in the remaining seven family members. This novel mutation generates a stop codon



The sequencing pattern for the family studied shows the hemizygous affected individuals with Kallmann syndrome, and heterozygous carriers who were detected by chromatogram patterns found in the study are shown above with mutations indicated by an arrow (B). The family members are given by I, II and III (A).