**Table 3**

Endocrine, Neuroradiological and extrapituitary manifestations of mutations in genes implicated in CCH in humans. E, Enlarged; N, Normal; TSH thyroid-stimulating hormone, PRL; Prolactin, GH; Growth hormone. AR, Autosomal recessive; XL, X-linked, \*Females may also be affected.

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| --- | --- | --- | --- | --- | --- |
| Gene | Inheritance | Hormone Deficits | TRH Test Responses | MRI | Additional Features |
| TSHB | AR | TSH | Absent TSH response, preserved PRL response | E, N | - |
| TRHR | AR | TSH | TSH & PRL peak absent/preserved | N | - |
| TBL1X | XL\* | TSH | TSH response normal | N | Sensorineural hearing loss |
| Isolated TSH Deficiency or combined pituitary hormone deficiency | | | | | |
| IGSF1 | XL\* | TSH ± PRL,  GH (transient)  Delayed pubertal testosterone rise | Low normal/Normal TSH response | N | Macroorchidism (males)  Ovarian cysts (females) |