Association between Congenital Hypopituitarism and Agenesis of the Internal Carotid Artery: a case report

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Background
The abnormalities of the Internal Carotid Artery (ICA) are very rare phenomena and the agenesis, in particular, has an estimated incidence of 0.01% among the general population. It could be also associated with another rare condition: the congenital hypopituitarism.

Method
Here we describe the case of a 5 months old female that presented respiratory distress after the birth and 1 month later, because of prolonged jaundice, was found to have low FT3, FT4, and TSH (FT3 was <2.3 pmol/L, FT4 was <3.5 pmol/L, TSH which was <0.1 microUI/L). She also had an undetectable Cortisol (<9 nmol/L) and IGF1 (<3.3 mmol/L).

MRI imaging demonstrated an ectopic posterior pituitary gland and the anterior pituitary gland appeared very small/hypoplastic. It also showed absence of the right ICA with an anastomotic vessel arising from the cavernous segment of the left ICA, crossing the midline, reconstituting the terminal right ICA and forming the right Middle Carotid Artery further on.

Conclusion
The case report we described is about a 5 months old patient with congenital hypopituitarism secondary to a hypoplasia of the pituitary gland, an ectopic posterior pituitary gland and the agenesis of the ICA.
Up to now this is the 14° case with the association between congenital hypopituitarism and abnormalities of the ICA and the 4° one before the first year of age.
Considering that a reduction of the blood supply is very unlikely to be the cause of the hypopituitarism, the hypothesis of a new, unknown, genetic mutation that could have caused both the pituitary hypoplasia and the agenesis of the ICA seems to be more likely.

References

I have no conflict of interest to disclose