Persisting Embryonal Infundibular Recess in a patient with Morning Glory Syndrome and multiple pituitary deficiencies

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Background

Morning glory syndrome is an association of morning glory disc anomaly (MGDA), a congenital optic disc defect, with other congenital abnormalities including , besides optic pathway,corpus callosum, craniofacial and skull base alterations, also vascular and pitiutary defects. The “persistence of embryonal infundibolar recesse (PEIR)” is a rare condition characterized by the lack of obliteration during development of a funnel-shaped liquoral space due to an expansion of the third ventricle floor into the pituitary stalk.

We present a case of a new rare association between MGDA and PEIR, previously not reported.

Case presentation

A 5-year old boy was referred to our clinic for short stature reported since first years of life. At birth weight and length were normal, psychomotor development was regular, target height was 165.9 cm (-1.68 DS). At 7 months of life he was subjected to correction of cleft lip-palate. Since 3 years of life he suffered from headache, for which a fundoscopy was performed and revealed a Morning Glory Disc Anomaly (MGDA) of the right eye.

At our first visit height was 98.2 cm (-2.5DS), body proportions were regular, IGF1 levels were low, bone age was delayed (3 years and 8/10). Stimulation test for GH secretion revealed a GH deficiency (arginine peak 2.2 ng/ml, glucagon peak 6.9 ng/ml).

MR imaging showed hypophyseal hypoplasia and a stubby, thickened, and inferiorly dropped optic chiasm with normal signal intensity. In sagittal images was also noted a dysmorphic hypothalamic infundibulum and pituitary stalk. Interestingly we found a direct communication between the third ventricle and the sellar cavity, suggesting a Persisting Embryonal Infundibular Recess (PEIR), the absence of sphenoidal meningocele was carefully proven . The sella was mildly enlarged, clival hypoplasia was noted. Additional findings were a corpus callosum body and splenium partial agenesis.

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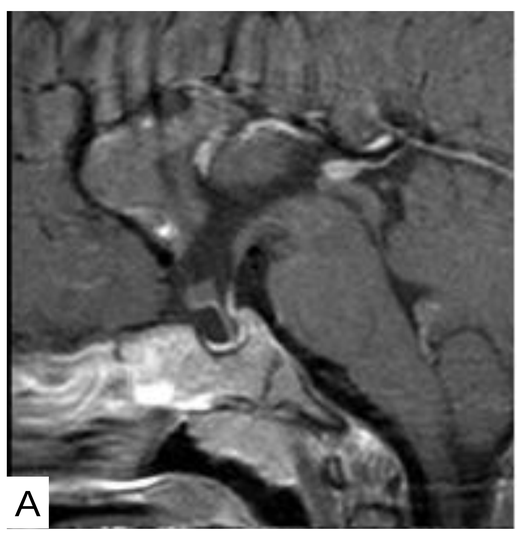


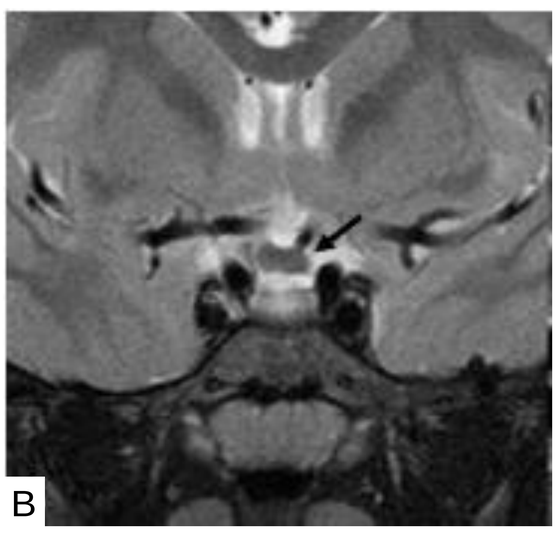
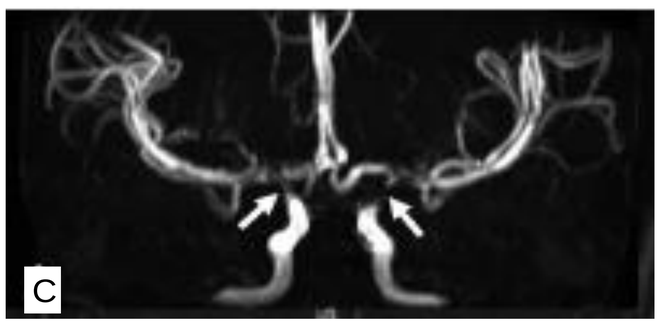


Figure A. MRI sagittal T1weighted image of patient
Figure B. MRI coronal T2-weighted image, black arrows indicate thickening of the optic chiasm

The Magnetic Resonance Angiography detected bilateral supraclinoid ICA (intracranial internal carotid artery) and M1segment of MCA (middle cerebral artery) narrowing, with thin collateral lenticulostriate vessels, compatible with a Moyamoya syndrome, the Perfusion Magnetic Resonance Imaging (DSC PWI study) revealed a preserved cerebrovascular reserve capacity.

During follow-up the patient developed also central hypothyroidism. Target gene sequencing of genes involved in hypopituitarism (*Gli2, Gli3, HESX1, LHX3, LHX4,OTX2, POU1F1, PROP1, SHH, SIX3, SOX3, TGIF, ZIC2*) and arrayCGH resulted both negative. Also *PAX6* gene mutations were excluded. We are planning a whole exome sequencing.



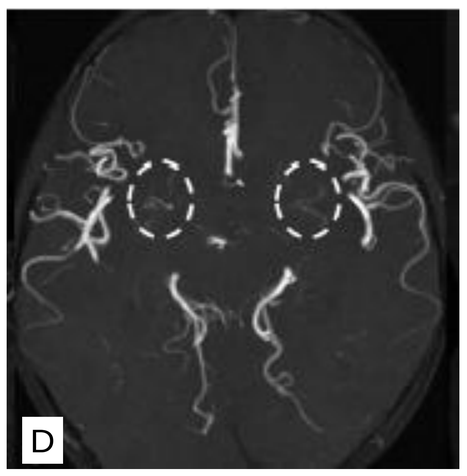


Figure C. MR angiography: bilateral focal stenosis of the internal carotid arteries;
Figure D. MR angiography: thin collateral lenticulostriates vessels

Conclusions

We described a complex case of Morning Glory Syndrome including pituitary and corpus callosum anomalies, Moyamoya syndrome, with a rare new association with Persisting Embryonal Infundibular Recess, this information may be useful in neuroradiological evaluation for the correct interpretation of an apparently duplicated pituitary stalk on coronal images

Adequate follow-up is required in patients with midline anomalies and MGDA to look for pituitary deficiencies and vascular abnormalities.

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No conflicts of interesse to declare.