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 pituitary defects. The “persistence of embryonal infundibular recesses (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 104 cm (-1.6SD). 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, IGFl 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.

The Magnetic Resonance Angiography detected bilateral supracilioid ICA (intracranial internal carotid artery) and Mlsegment of MCA (middle cerebral artery) narrowing, with thin collateral lenticulostrate 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 (GIL1, GIL3, HESX1, LHX3, LH4X, OTX2, POU1F1, PROP1, SHH, SIX3, SOX3, TGF, ZIC2) and arrayCGH resulted both negative. Also PAX6 gene mutations were excluded. We are planning a whole exome sequencing.

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.

References


No conflicts of interest to declare.