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Agenesis of septum pellucidum with hypophyseal abnormalities – case report

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Introduction:
Agenesis of septum pellucidum is a rare congenital anomaly of the newborn with an incidence of 2-3 per 100 000 children. There is tremendous phenotypic heterogenicity and an unknown genetic reason (<10% of the cases have associated gene). The condition is sometimes part of a syndrome called septo-optic dysplasia (SOD) that is characterized by hypoplasia of the optic nerve, hypophyseal insufficiency and structural defects in the midline of the brain (different anomalies in septum pellucidum and/or corpus callosum). Prenatally the anomaly can be assumed when there is hypo- or dysgenesis of the septal leaflet, which leads to complete lack of visibility of the cavum septum pellucidi. This finding has vast differential diagnosis – holoprosencephaly, agenesis/dysgenesis of corpus callosum, hydrocephalus etc. In these cases the recommendation is to obtain prenatal magnetic resonance imaging (MRI) or continue supervision after birth with ultrasound and if needed MRI. Vision and endocrine function of the hypophysis can be assessed only postnatally. Clinical presentation can be diverse, the main symptom is impaired vision (because this condition is mainly part of SOD syndrome), followed by anomalies of the pallatum durum, hypopituitarism with predominant involvement of somatotropin and different neurological injuries (paralysis, paresis, seizures, nystagmus etc.). Prognosis is unclear - should be discussed on an individual basis depending on the severity of organ involvement. Treatment includes multidisciplinary management which can include hormone substitution, anticonvulsants, physio- and kinesitherapy, visual impairment is generally untreatable.

Discussion:
Agenesis of septum pellucidum is a rare condition with multiform presentation, in a lot of the cases concomitantly with hypoplasia of the optic nerve as a part of SOD. Our patient has isolated septum anomaly with hypopituitarism without any pathology from the optic nerves. In some cases the anomaly is recognized prenatally during routine fatal morphology examinations, but in this case the condition was not recognized. Abnormal development of brain structures can produce different neurological symptoms with varying severity – seizures, paresis, paralysis etc. Our patient had multiple apnoic pauses and reduced muscle tone with normal motor function as well as normal, symmetrical reflexes of the newborn. Hypopituitarism can range from panhypopituitarism to isolated injury of a single endocrine function and required hormone substitutes. The presented case has hypocorticism and hypothyroidism without episodes of hypoglycemia. Additionally this patient will require strict growth measurements and in the future thorough assessment of pubertal development. Long term prognosis is unclear and depends on the symptoms that have developed. Cases with severe neurological involvement and those with ocular involvement have the tendency to be with retardation in the neuropsychological development. Our patient has mild neurological involvement that became better after hormone substitution and has better prognosis than other cases.

Conclusion:
Agenesis of septum pellucidum is a rare condition with polymorphic symptoms, hard to assess and diagnose in the neonatal period. Careful physical examination with the use of modern imaging studies helps in quickly distinguishing the disease, evaluating it and starting the appropriate treatment.

Biography
Dr Shteryu Boyadzhiev is currently working as a Senior Consultant Neonatologist and Head of Neonatal Intensive Care Unit (NICU) at University Hospital Lozenetz Hospital, Sofia, Bulgaria. Also worked as a Honorary Senior Lecturer at Sofia University "St.Kliment Ohridsky" - Faculty of Medicine.

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