Pituitary aplasia - A rare cause of neonatal hypoglycaemia

Clinical History:

A full term male newborn presented with hypoglycaemic convulsions and neonatal hyperbilirubinaemia. Birth weight was normal. Examination at birth revealed single umbilical artery and micropenis, but no other anomalies were present.

Imaging Findings:

Brain MRI of the baby at 1 ½ months age revealed absent anterior lobe of pituitary gland and absent posterior pituitary bright spot with non-visualisation of the pituitary stalk. The sella turcica was shallow and flattened. The supra and parasellar structures were normal and no structural abnormality or migrational defects were seen.

Discussion:

Aplasia or absence of pituitary gland is a rare congenital anomaly which presents with consequences of multiple pituitary hormone deficiency and involves absence of anterior and posterior pituitary gland and the pituitary stalk [1, 2].

Neonatal hypoglycaemia is common, but when associated with microphallus or micropenis [3] should raise the suspicion of congenital hypopituitarism and prompt endocrinologic assessment and brain MR imaging [1, 4]. Deficiency of testosterone or growth hormone may be associated with micropenis as these hormones are essential for penile growth beyond 14 weeks of gestation [1].

Neonatal hypoglycaemia can present in the first few hours of life manifesting with seizures, apnoea and cardiovascular collapse and respiratory distress [1, 4], hypotonia and non cholestatic jaundice [5]. These infants are usually delivered at term and have normal birth weight; however, some anomalies such as micropenis, small or undescended testes or poorly developed scrotum may be present. Some may have associated midline facial anomalies [2, 3] like cleft palate and lip, hyper/hypotelorism and deviated nasal septum. Growth retardation generally presents after 6-8 weeks, but at birth the newborn is of normal growth due to maternal growth hormone [1].

Hypopituitarism due to pituitary aplasia in newborns may present as an isolated anomaly or may be associated with craniofacial defects, absent septum pellucidum, anencephaly, arrhinencephaly, holoprosencephaly and septo-optic dysplasia [1, 3]. Hypoglycaemia in these neonates may be due to cortisol deficiency as a result of reduced adrenocorticotropic
hormone and growth hormone. As the child grows, cortisol replacement is needed for glucose control as the adrenal cortex in these infants is generally absent or severely atrophic [1, 3]. These infants do well when multiple pituitary hormone supplementation especially cortisol and thyroxine is initiated early [4, 5].

MRI of the brain in these newborns shows complete absence of adeno and neurohypophysis along with the pituitary stalk and associated shallow sella turcica [1, 2]. Hypoplasia of the pituitary gland is a differential diagnosis and can be differentiated from aplasia by presence of a pituitary stalk, ectopic posterior pituitary and a well formed sella. Another differential for non-visualised pituitary is the empty sella syndrome, where the sella is well formed and pituitary functions are normal and less commonly seen in children [1].

**Differential Diagnosis List:** Pituitary aplasia, Pituitary hypoplasia 1, Empty sella syndrome 1

**Final Diagnosis:** Pituitary aplasia

**References:**


Description: T1W Sagittal MRI shows non-visualisation of pituitary gland, pituitary stalk and posterior pituitary bright spot. 

Origin: Department of Radiodiagnosis, Father Muller Medical College Hospital, Mangalore, India.
Figure 2

Description: T2W coronal MRI shows absence of pituitary gland and stalk and also a shallow sella.
Origin: Department of Radiodiagnosis, Father Muller Medical College Hospital, Mangalore, India.