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IMAGES IN CLINICAL PRACTICE

# DIENCEPHALIC CACHEXIA - RARE YET CRUCIAL ENTITY

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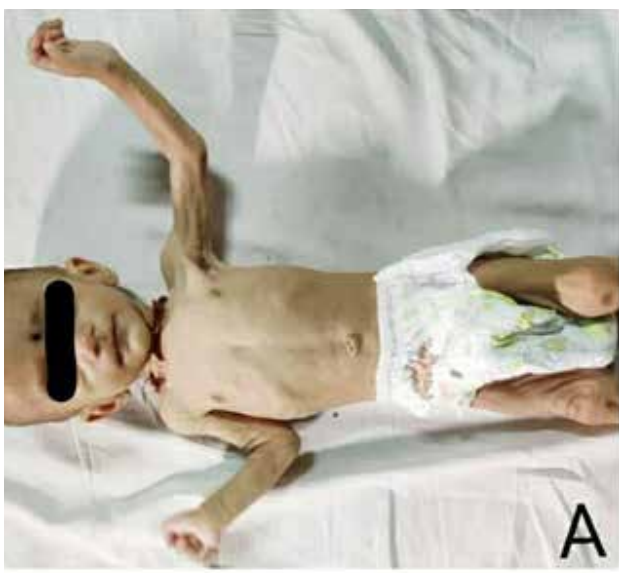
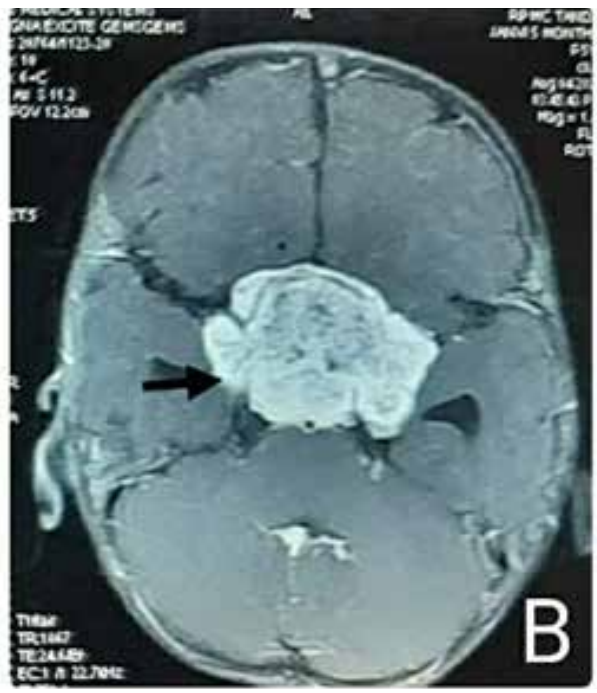
A 5 months old female infant born of non-consanguineous marriage presented with complaints of not gaining weight (<3rdpercentile) for 3 months despite adequate calorie intake and continuous abnormal eye movements for 1 month. This infant was a full term born by vaginal delivery with birth weight of 2.5 kg. On examination vitals were stable. Infant looked emaciated and hyperactive (Fig.1 A). The nervous system examination revealed profound muscle wasting in all 4 limbs, generalised hypertonia with brisk deep tendon reflexes. Eyes showed continuous horizontal nystagmus in every gaze. Other systems were normal. MRI brain with contrast showed well defined lobulated mass measuring 3×2.6×4.4 cms in size in suprasellar region arising from hypothalamus (Fig.1 B,C,D) with possibility of glioma. Patient is planned for surgical intervention with the final diagnosis of Diencephalic syndrome (DES).

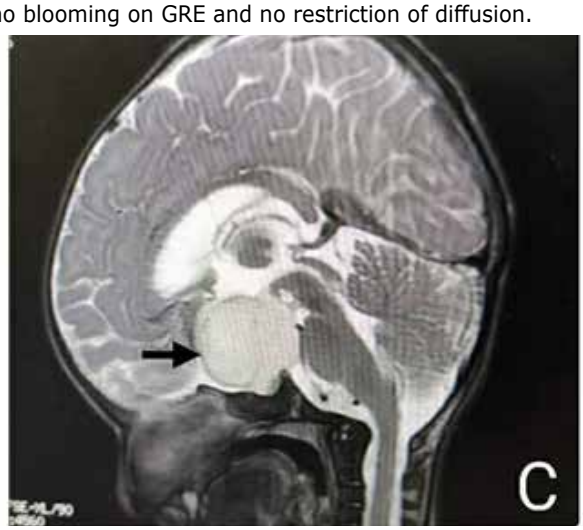
**Figure 1.** Diencephalic Syndrome
**A:** Infant with faltering of growth




**B:** MRI of Brain-T1 Axial section showing lobulated mass which is homogenously hypointense in suprasellar region arising from hypothalamus with intrasellar extension.




**C:** MRI of Brain-Sagittal section showing lobulated mass hyperintense on T2 extending into 3rd ventricle and invading bilateral thalamus, with avid post contrast enhancement, no blooming on GRE and no restriction of diffusion.

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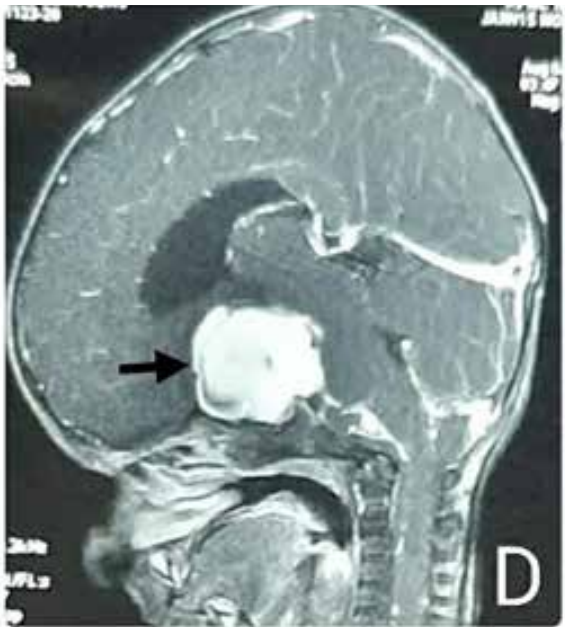
**D:** MRI of Brain-T1 Sagittal section showing lobulated mass in suprasellar region arising from hypothalamus with intrasellar extension.




*What is diencephalic cachexia?*

Diencephalicor Russell's syndrome is characterized by profound emaciation during infancy despite normal or slightly reduced calorie intake, nystagmus and hyperkinesis due to intracranial neoplasm involving anterior hypothalamus or optico-chiasmatic glioma[1] This article emphasizes on an exceptional yet significant reason for cachexia which often leads to delayed diagnosis.

Diagnosis of DES is based on the major criteria comprising of emaciation, locomotor hyperactivity, euphoria and minor criteria which are skin pallor, hypotension and hypoglycemia.[2,3] Pathogenesis of weight loss include the hypothesis of elevated growth hormone (GH) level with paradoxical response to a glucose load, partial GH resistance and excess of ß-lipotropin (lipolytic peptide produced in excess by primary neoplasm or due to secondary effect of invasion), resulting into fat breakdown and complete loss of subcutaneous adipose tissue.[4] DES occurs due to mass effect caused by glioma in the sellar–suprasellar hypothalamic region. This also results into decreased visual acuity and visual fields in 65% of patients. MRI remains the diagnostic modality while surgery is the mainstay of treatment with adjuvant chemotherapy and radiotherapy. Pediatricians must keep DES as one of the differential diagnosis of unexplained inability to gain weight.

**Contributors' Statement**

SS, SK, and AS conceptualized the perspective, drafted the manuscript. SS, Isha, and KS reviewed and revised the initial manuscript. All authors approved the final manuscript as submitted and agree to be accountable for all aspects of the work.

**Compliance with ethical standards**

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