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Case Report

A rare case of congenital aqueductal stenosis

Col K.S. Brar^{a,*}, Brig A.K. Sharma^b, Surg Lt Cdr Ajay Nathan^c^a Senior Advisor (Medicine & Endocrinology), Army Hospital (R&R), Delhi Cantt.- 10, India^b Consultant (Surgery & Neuro-surgery), Department of Neuro-surgery, Army Hospital (R&R), Delhi Cantt-10, India^c Graded Specialist Medicine, Military Hospital, Shimla, India

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Introduction

The authors present a case of severe short stature with delayed puberty in a 22-year-old patient found to have non-communicating hydrocephalus due to non-tumoral aqueductal stenosis. Although the presentation of hydrocephalus with delayed puberty and short stature has been previously reported, this association remains rare.¹ Also concomitant Growth Hormone deficiency (GHD) and central hypothyroidism due to hydrocephalus is extremely rare. This is most likely being reported for the first time in this case report. After treatment with V-P Shunt, the patient has shown increase in height, entered puberty spontaneously but continues to require L-thyroxine (LT4) therapy. A review of the literature on the endocrinological disturbances in patients with hydrocephalus is presented.

Case report

22-year-male patient presented with complaints of not attaining secondary sexual characters, in the form of not having beard and pubic hair and also complained of small penile length. He denied any other symptoms. He had finished his college with below average grades and was working as farmer with his father. On examination he had height was 140.7 cm (below 2 SD), Mid Parental Height (MPH) 160 cm, Arm span 139 cm, round chubby face, Weight 52 kg, Body Mass Index (BMI) 26 kg/sq m, absent facial, axillary and pubic hair (Tanner stage 1), Stretched Penile Length (SPL)-5 cm, Testicular volume 2 mL bilateral and had lipomastia (Fig. 1). IQ was 88. Rest general and systemic examination was essentially normal.

Investigations revealed normal hemogram and routine biochemistry including Hemoglobin, Creatinine and ESR. Endocrine evaluation revealed: TSH – 0.42 μ IU/mL, Free T4 – 0.70 ng/dL, Basal GH – 1.8 ng/mL, Basal serum Cortisol – 13.6 μ g/dL. Serum Testosterone level – 1.2 nmol/L. Serum LH 1.2 U/L, FSH – 2.1 U/L. Serum Prolactin – 24 ng/mL. Peak GH Level with Clonidine stimulation was 2.2 ng/mL. Insulin Induced hypoglycemia test was carried out and samples collected for GH, TSH and Cortisol during hypoglycemia which showed peak GH level 2.8 ng/mL, TSH – 0.50 μ IU/mL, Cortisol – 18.4 μ g/dL. X-ray Lt hand including wrist joint showed bone age of 17.1 years and epiphysis were still not fused (Fig. 2). MRI Brain revealed large hydrocephalus with entire pituitary flattened against the sella wall (Fig. 3).

Patient was started on LT4 replacement. He was taken up for ventriculostomy but due to unfavorable anatomy (Floor of 3rd ventricle couldn't be visualized) he underwent V-P Shunt for congenital aqueductal stenosis which was causing hydrocephalus.

* Corresponding author. Tel: +91 8017298837.

E-mail address: ksbrar32@hotmail.com (K.S. Brar).

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Fig. 1 – Photograph of the patient, 22-year-old male.

Patient was started on Growth Hormone and LT4 therapy continued. Testosterone replacement was not initiated due to still possible potential for gain in height which could be adversely affected by androgen replacement. Patient after 07 month felt subjectively better and more active, he gained 4.2 cm height and lost 4.5 kg weight. Testicular volume Rt- 4 ml, Lt-5 ml, developed pubic hair (Tanner stage 2). TSH – 1.0 μ IU/ml, Free T4 – 1.3 ng/dL, Serum Testosterone level – 5.2 ng/mL, Serum PRL – 12.0 ng/mL. Repeat MRI brain revealed partially decompressed ventricles with decreased pituitary flattening in sella and visualization of normal posterior pituitary bright spot (Fig. 4). He is under regular follow-up.

Fig. 2 – X-ray Lt hand including wrist joint (AP view) markedly delayed bone age 17.1 years (TW3).

Fig. 3 – MRI of Brain, pituitary sections (Sagittal section) showing large hydrocephalus with entire pituitary flattened against the sella wall.

Discussion

The word “hydrocephalus” is derived from two Greek words: hydro meaning water; and cephalus meaning head; also known as “water on the brain”. Historically it is believed to

Fig. 4 – Repeat MRI of pituitary (Sagittal section) (03 months Post-operative) showing partially decompressed ventricles with decreased pituitary flattening in sella and visualization of normal posterior pituitary bright spot.

result from imbalance between CSF production and absorption, with net accumulation of fluid in the cranial cavity; characterized by increase in size of the cerebral ventricles. It is classified as: Communicating hydrocephalus, in which flow is not obstructed, but CSF is inadequately reabsorbed in the subarachnoid space and the Non-communicating hydrocephalus or the Obstructive type, in which flow of CSF from the ventricles to subarachnoid space is obstructed. This type may also be sub-classified into Congenital and Acquired. The overall incidence of hydrocephalus is not known. Approximately 55% of all hydrocephalus are congenital.² The etiology depends upon the age of the child. The clinical features are increase in the size of head, with wide anterior fontanelle, prominent scalp veins, sun-setting eyes, optic nerve atrophy, nystagmus and increased muscle tone in children upto 2 years. Children more than 2 years may present with these as hydrocephalus progresses; or if the fontanelle is closed, head size may be normal. These may present with optic atrophy or papilledema, abnormal hypothalamic functions (short stature or gigantism, obesity, early or delayed puberty, primary amenorrhea or menstrual irregularity and diabetes insipidus) and spastic lower limbs. Performance IQ is worse than verbal IQ and learning problems are common. Hydrocephalus associated with aqueductal stenosis is not uncommon after the 2nd year of life.³ Intracranial hypertension is the most frequent symptom at diagnosis. In patients with aqueductal stenosis, endocrine dysfunctions are evident in 25–35% of the cases.⁴ Of the endocrine dysfunctions most frequent dysfunction is Central early puberty (25–50%), followed by GH deficiency (10–30%) or neurosecretory GH dysfunction.¹ Both hormonal disorders may be the consequence of damage to the hypothalamus or the pituitary gland caused by increased intracerebral pressure or increased mass of cerebrospinal fluid. Free thyroxine is abnormally low in 3.2% patients only. Cortisol is almost always within the normal range in these patients.⁵ In children with hydrocephalus, short stature is rare and is frequently due to meningomyelocele and very rarely to GH deficiency.¹ Our patient has rare presentation of severe

delayed puberty and short stature due to concomitant central hypothyroidism and GH deficiency, detected in 3rd decade of life. To our knowledge this is the first reported case of concomitant central hypothyroidism and GH deficiency in a patient with congenital hydrocephalus. The patient has responded well to L-thyroxine, GH therapy and surgical procedure of intracranial decompression by V-P Shunt. Delayed puberty in non-communicating hydrocephalous is also rare as these patients usually present with hypothalamic central precocious puberty. Central hypothyroidism seen in this is a very rare endocrine abnormality in patient with long standing hydrocephalous and requires L-thyroxine replacement even after surgical decompression. This case is being presented due to its rarity.

Conflicts of interest

All authors have none to declare.

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