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Pineal Body Tumour, A Very Rare but Often Misdiagnosed in Our Clinics and Literature Review

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ABSTRACT: This publication aims to review and remind Medical Practitioners of a very rare and often misdiagnosed case of a central nervous system tumour, pineal body tumour. A 4-year old boy child referred from a peripheral hospital in a neighbouring town on account of not responding to several treatments. Mother booked for antenatal care by 3rd month of the pregnancy. She adhered to her treatments and kept to her appointments throughout the care. Mother had a hitch-free labour and delivery. Immediate neonatal and postnatal life was good up to the onset of the present illness. Pineal body tumour is a very rare and often misdiagnosed malignancy because of its several mimicries, which demands a very high index of suspicion and extensive clinical assessment to diagnose. Presently there is daft information on this rare brain tumour, and so this publication.

KEYWORDS: Pineal gland, hydrocephalus, epithalamus

PRESENTATION

K.C is a four-year old boy child brought to our clinic by her aunt, as his mother was said to be nursing a newborn baby. They came from a peripheral hospital in a neighbouring town on account of K.C's worsening health condition despite being on treatment in the hospital under Doctor's care. She said the child has been on admission for more than three weeks at the said peripheral hospital, and his health condition was worsening rather than getting better. Therefore, they requested for discharge and were referred on demand to a higher level of care.

At the peripheral hospital, their complaints included pain in the head (headache), poor feeding, and poor sleep. He was, after assessment and laboratory investigations, admitted and commenced on treatment for severe malaria and enteric fever with parenteral anti-malaria and antibiotics regimen respectively. As the days went by while on admission receiving above treatments, the child started complaining of visual disturbances with attendant poor movement, bumping unto objects and persons with frequent falls. With further deterioration of his general health condition including his vision, the child started shouting that he could no longer see anything. At this point, the patient's relations requested for discharge against medical advice (DAMA), which it was said the Doctor granted with grudges. The child was then brought to our clinic.

On presentation to our clinic, the following information was obtained from K.C's aunt: prior to the present illness K.C has remained in good state of health since his birth 4 years ago. His aunt, amongst the above complaints, also noticed increase in his head size, limbs becoming stiff with loss of walking and sitting abilities. K.C also developed several episodes of convulsions, each of tonic-clonic in form, but could not estimate the duration of any episode or if they were preceded by aura.

The mother's pregnancy of the index child was said to be planned and achieved. Booked for antenatal care (ANC) by the third month of the pregnancy, and was regular with her appointments including her immunization schedules. No history of exposure to radiation or unusual meals including carcinogenic agents during the care. Pregnancy was said to be carried to term, labour not prolonged and child cried immediately after birth. No immediate and post-natal problem and early childhood life said to be good till the onset of the present health challenges.

Patient's immunization was incomplete with no cogent reasons adduced for the missed ones.

K.C is the first child of a monogamous family with 3 children: 1st is the index child, K.C, 2nd is a two(2+) - year old boy child who is alive and well, while the 3rd is a two-month old baby girl who is also alive and well. No similar ailment has been noted in the family lineages. Mother is a 32-year old graduate who is self-employed while the father is a 40-year old West African School Certificate (High School) holder who deals on groceries and beverages. K.C lives with his biological parents in a two-room apartment with good ventilation.

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On examination,
patient was conscious but irritable, moderately nourished, afebrile, mildly anaemic, not dehydrated, nil peripheral oedema and nil palpable lymphadenopathy of significance. BW 16 kg, Temp 37.2^{oC}, PR 80 cpm, RR 22 cpm, and BP 86/60 mmHg.
Nervous system examination revealed a conscious but irritable boy, well dressed with head size bigger for age (OFC 52 mm). Also in mild opisthotonic posture, fontanelle closed, no prominent peripheral veins over the skull and cranial sutures not separated (intact).

Spastic quardriplegic limbs, with plantar flexion of both feet, Babinski positive on limbs, clonus sustained and muscle bulk intact.

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Ophthalmic findings include limitations of extra-ocular movements in all directions of gaze. There was loss of vision in both eyes, nil megacolonic eyeballs (not proptic), globes were mildly sunset; pupils were mid-dilated and reacted sluggishly to light stimulus. Fundoscopy revealed bilateral papilloedema and retinal vasculitis.

Respiratory and digestive systems were of normal findings. There is no visceral organomegaly.

Diagnosis of Space Occupying Lesion (SOL) ? Cause, was made;

Keep in view(KIV): Intracranial tumours(ICT).

Urgent computerized tomographic scan (CTS) request was done and the result revealed Pineal body/gland tumour.

Patient was admitted and Neurosurgeon invited for further evaluation and subsequent management vis-a vis resection of the tumour mass. Presently, K.C is under a multidisciplinary care involving the Paediatrician, Ophthalmologist and Neurosurgeon and being worked up for surgery and subsequent chemotherapy.

Introduction: Pineal body/gland is a pinecone-shaped small gland, measuring 0.3'' long and weighs about one-tenth of a gram weight (0.1g), located in the middle of human brain in between the hemispheres in an area medically known as epithalamus.[1] It is composed of pinealocytes and supporting cells, equivalent to brain astrocytes. The pineal region, according to Ringertz et al,[2] defined the pineal region as being bound by the splenium of the corpus callosum and tela choroidea dorsally, the quadrigeminal plate and midbrain tectum ventrally, the posterior aspect of the third ventricle rostrally, and the cerebellar vermis caudally.

Its functions are regulated by the innervating adrenergic nerves. Its main hormones are melatonin and serotonin. The later is the precursor of the former, melatonin, which regulates the body's "Circadian rhythm (sleep-wake patterns). Pineal gland hormones

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also impacts positively on the health of cardiovascular and reproductive systems [1]. Its anatomical location in the peri-aqueductal region in the brain results, with any increase in its size, to compression of adjacent structures with the resultant ocular manifestations[2].

Pineal Body Tumour, PBT is a very rare clinical entity that is seldom seen in Paediatrics, accounting for 0.5% of all central nervous system tumours in adults, 1% in young adults(aged 20-34years), and 2.7% in children (aged 1-12years) [1]. Because of the rarity of the tumour, it has been difficult to collect a large number of cases to study and compare [1, 3].

Aetiology of the tumour is unknown, as is mostly the case with other childhood malignancies [1].

DISCUSSION

Pineal body tumour (PBT) clinical features at early onset mimic other ailments in our environment including infective processes [1,4,5].

K.C initially presented with headache, a non-specific symptom that can be seen in many other mimicking ailments including malaria and enteric fever in the tropics. Headache is one of the common features of PBT as was observed in the index patient.

Our patient complained of loss of appetite throughout his illness that never improved with all the management interventions. This may not be too strange in the context of the patient's morbidity – PBT – that is well associated with nausea and loss of feeding [1,2]. In our environment also, loss of appetite is common with enteric fever, malaria and septic illnesses.

The classic triad of headache, nausea and vomiting, and papilloedema is associated with midline or infratentorial tumours as seen in this patient with PBT [1, 2]. This triad may also be seen in the tropics in infective processes like meningitis, encephalitis, etc.

In one of his earlier presentations, K.C complained of poor sleep, both during the day and at night. This is one of the pathophysiological features of PBT where disruption in sleep patterns is a norm. [1] This complaint follows the disruption of the 'Circadian rhythm' regulation by its hormone – melatonin.

Jeffrey N Bruce et al, in their study [4], stated that clinical syndromes associated with pineal region tumors relate directly to normal pineal anatomy. According to their findings, lesions in the pineal region will lead to aqueductal compression with resultant hydrocephalus. This study finding collaborates with one of the index patient's presenting complaints, increased head size. The study also stated that further compression of the peri-aqueductal gray region may cause mydriasis, pupillary inequality (anisocoria) and papilloedema. These worsening ocular features were also found in this our patient as his ophthalmic assessment revealed mid-dilated pupils that reacted sluggishly to light stimulus.

The ocular findings associated with Pineal Body Tumors result mainly from its anatomical location in the peri-aqueductal region in the brain, leading to compression of adjacent structures with the resultant ocular manifestations[2].

Other authors have highlighted the different ocular findings in patients with pineal body tumors [5,6, 7 etc].

Harkinson EV, in a particular study [5], found papilloedema in 69% of patients. This study finding collaborates with the fundoscopic finding in the index patient that revealed bilateral papilloedema and retinal vasculitis.

In another report by Akiko M et al [6], a 17-year old male patient with pineal germinoma presented with bilateral blurred vision, bilateral uveitis, bilateral retinal periphlebitis (juvenile retinal vasculitis), a proliferative membrane extending from the papilla to the macula and optic neuritis with observed tractional serous retinal detachment around the macula. The study's later findings collaborate with our patient's ocular findings. In contrast, the earlier ocular findings of this study were not found in our patient due his late presentation to us as well as his age, 4 years against the 17 year-boy in the Akiko M et al's study.

In that medically reviewed article by Davis 2022 [3], he reported that pineal body tumours may cause visual changes with associated symptoms and signs which may include inability to focus on objects, double vision, and impairment of eye movements. These study findings may be possible in early presentations as the index patient has lost his vision before presenting to our clinic.

Pineal Gland Tumour treatment options: The first treatment for pineal region tumour is *surgery*, if possible. The goal of surgery is to obtain tissue to determine the tumour type/grade and to remove as much tumour tissues as possible without causing more symptoms for the patient. Treatments after surgery may include *radiation, chemotherapy, or clinical trials* [5].

Pineal Gland Tumours prognosis: The relative 5-year survival rate for pineal region tumours is 69.5% but it is known that many factors can affect prognosis. These include the tumour grade and type, traits of the cancer, the person's age and health when diagnosed, and how they respond to treatment [5].

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PBT is a very rare clinical entity that is seldom seen in Paediatrics practice. The rarity of the tumor can be improved upon by high index of suspicion as well as availability of supporting diagnostic tools. This is the case in the index patient where a very high level of suspicion was taken and ably supported and clarified by available diagnostic facility.

Because of the rarity of the tumour, PBT, there is the high probability of missing this tumour when it presents in the clinic and worse in its early stage when most of its symptoms mimic those of other common illnesses in our environment. In view of this, we need to re-emphasize here the need for sound clinical exploration of patients' symptoms and signs.

Conclusively, the rarity of Pineal Gland Tumour, PGT, as well as its features mimicking features of other prevailing illnesses in our environment makes it a case of high possible misdiagnosis.

In view of this, this publication aims to remind us on the existing of this rare problem among us and encourage us to increase our index of suspicion of this rare illness, PBT. This is very important as we all know that early diagnosis and treatment of the tumour, like other tumours, reduces its attendant morbidities and mortalities.

This publication also hopes to reduce the wide gap created by daft publications on this rare but often misdiagnosed tumour, Pineal Body Tumour, PBT.

The political will of the government at all levels in improving health care services including equipping our laboratory units in our health facilities will also help in achieving the above feat.

With improved patient assessment and high index of suspicion we can do better.

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