INTERNATIONAL JOURNAL OF MEDICAL AND APPLIED SCIENCES
Earthjournals Publisher
E-ISSN:2320-3137
www.earthjournals.org

CASE REPORT

OCULAR MANIFESTATION OF COCKAYNE'S SYNDROME- CASE REPORT

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ABSTRACT:
The cockayne syndrome is rare autosomal recessive disorder presenting in early childhood. Growth failure, mental retardation and developmental delay are usually associated with charactacteristic facies, photosensitivity, microcephaly, retinal pigmentation and deafness. The limbs are disproportionately long with flexion contracture and kyphoscoliosis may develop later. Patients with Cockayne syndrome show extensive loss of vision by the second decade of life and have RP, optic atrophy, and cataracts dwarfism, deafness, mental deterioration, and premature aging.

In our case external eye examination showed that both eyes had deeply set eyes, enophthalmos due to marked loss of subcutaneous fat and orbital fat. Eyelid closure were normal. Bells phenomenon were normal. Conjunctiva and cornea was normal. There was no nystagmus. Patient had alternating concomitant exotropia of 60 prism diopters. Direct and indirect pupillary reaction was present and was sluggish in both eyes. Pupillary dilation was poor after instillation of tropicamide and homatropine eye drop. Lens was clear in both eyes. Lacrimal secretion was adequate.

Keywords : cockayne syndrome, autosomal, case report, ocular manifestation

INTRODUCTION

The cockayne syndrome is rare autosomal recessive disorder presenting in early childhood. Growth failure, mental retardation and developmental delay are usually associated with charactacteristic facies, photosensitivity, microcephaly, retinal pigmentation and deafness. The limbs are disproportionately long with flexion contracture and kyphoscoliosis may develop later. (3)

Patients with Cockayne syndrome show extensive loss of vision by the second decade of life and have RP, optic atrophy, and cataracts dwarfism, deafness, mental deterioration, and premature aging . (4, 5)

CASE REPORT:

An 11 yr. old male child referred for ophthalmic evaluation from paediatric department. He wasadmitted for developmental delay. He had hearing and speech delay. He was a full term

baby with birth wt. of 1.8 kg. Since birth was dwarf. On further evaluation it was found that he has microcephaly (head circumference 43 cm). He had prominent nose, shrunken eyes, loss of subcutaneous fat, senile look, bird like face, contractures, stiffness, height was 90 cm with proportionate short stature, scoliosis.{fig.1 2 3}. CT scan showed cerebral atrophy with calcification {fig.4}. BERA showing severe sensory neuraldeafness {fig. 5 and 6}.

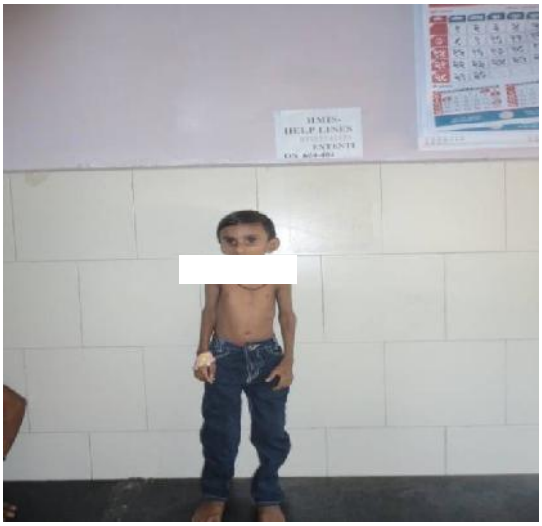

Figure 2

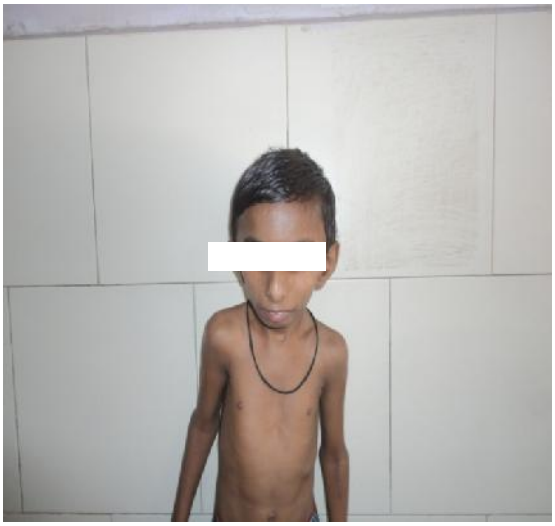

Figure 1

Figure 3

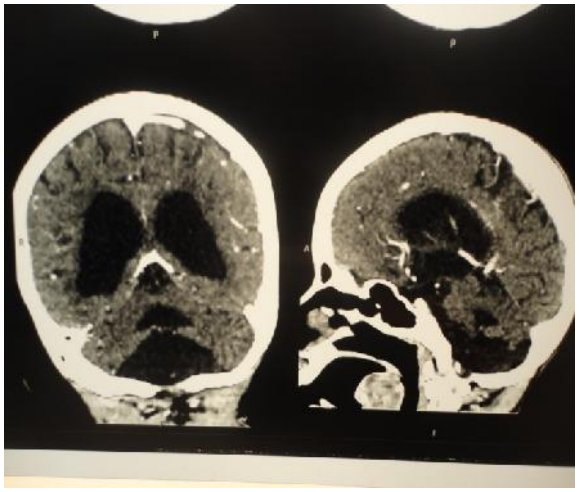

figure 4

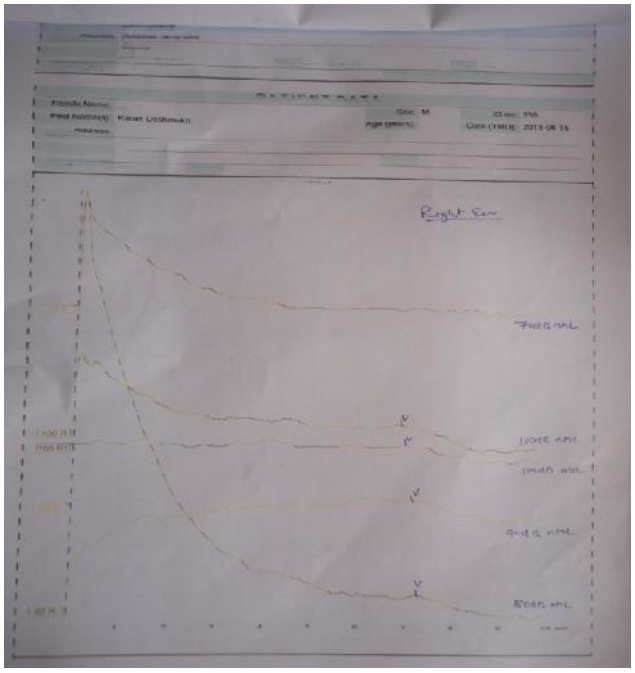

Figure 5 BERA right ear

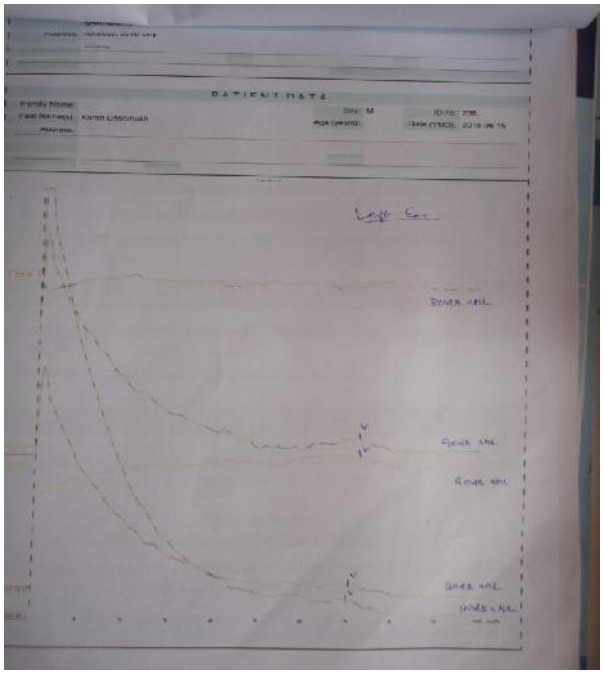

Figure 6 BERA left ear

Ocular manifestation:

External eye examination:

In our case external eye examination showed that both eyes had deeply set eyes, enophthalmos due to marked loss of subcutaneous fat and orbital fat. Eyelid closure were normal. Bells phenomenon were normal. Conjunctiva and cornea was normal. There was no nystagmus. Patient had alternating concomitant exotropia of 60 prism diopters. Direct and indirect pupillary reaction was present and was sluggish in both eyes. Pupillary dilation was poor after instillation of tropicamide and homatropine eye drop. Lens was clear in both eyes. Lacrimal secretion was adequate.

Visual acuity could only be estimated by observation and appeared to be about 6/60 in each eyes. Patient had extreme photophobia Mental retardation made determination of visual field not possible.

On slit lamp examination both cornea appeared normal in size and shape. There was no evidence of corneal erosion and fluorescein staining was negative. Anterior chamber was clear and irises normal in both eyes. No lens opacities were found. The vitreous was normal.

Fundus examination:

On fundus examination of both eyes patient had clear media with partial optic atrophy, fine retinal pigment mottling in periphery and posteriorpole. Foveal reflex was absent. Retinal arteries were attenuated and veins were normal.(Fig 7 & 8)

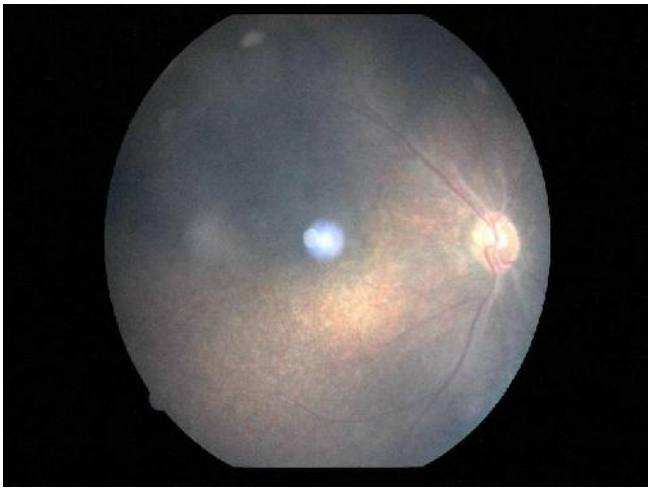

Figure 7 fundus photo right eye

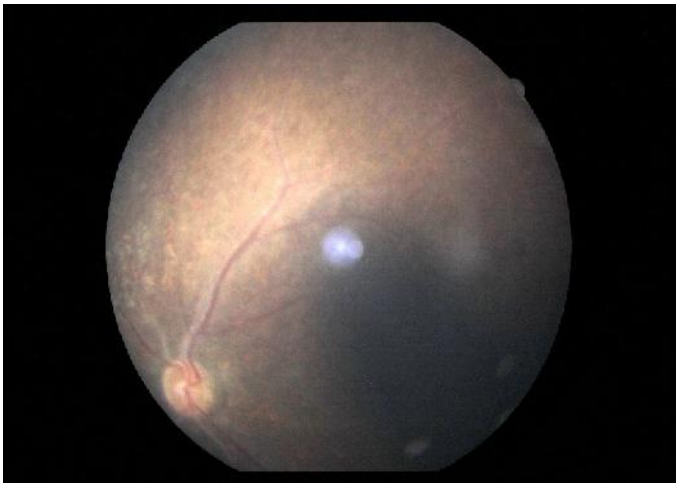

Figure 8 fundus photo left eye

Discussion:

In 1936,' and again in 1946, Cockayne described a syndrome of "dwarfism with retinal atrophy and deafness." Clinical features become evident at age 1 to 2 years and consist of cachectic and disproportionate dwarfism. (1, 2)Musculoskeletal changes include kyphoscoliosis, ankylosis, a horseriding stance, long limbs, and large hands and feet. (6, 7) Patients are microcephalic and have a progeroid facies with lack of subcutaneous fat, prominence of the facial. Bones, enophthalmos, a thin often beak-like nose, large ears, and dental abnormalities. Sensorineural deafness and progressive neurodegeneration with mental retardation, cerebellar ataxia, choreoathetosis, epilepsy, extrapyramidal tract signs, intracranial calcifications, and peripheral neuropathy become evident with time. (8)Photo dermatitis of sun-exposed areas is a prominent feature and was present in several of our patients. When the diagnosis is suspected, parents should be questioned about the presence of facial photosensitivity. Death ensues in the second to fourth decades. There appears to be an early-onset form of Cockayne syndrome with intrauterine onset of growth retardation and evidence of clinical features at birth. (9, 10, 11)

Enophthalmos in these patient is caused by loss of subcutaneous and orbital fat. (4) Poor dilatation in these patient is due to atrophy of dilator muscle fibers atrophy as evidenced by iris Trans illumination defect. (4). Cataracts have been described in about 15% of patients21 and were present in both of Cockayne's original patients. (1, 2)

Photoreceptor and iris damage in patients with Cockayne syndrome may be caused by ultraviolet light-induced DNA and RNA replication defects. In the absence of effective recovery of DNA and RNA replication, cells that receive large amounts of ultraviolet light degenerate, leading to the clinical ocular features. Additionally, because of the high metabolic activity in photoreceptor and retinal pigment epithelial cells, selective loss of repair of transcriptionally active DNA may result in more damage to these cells as postulated for neurons by Venema and associates. (12)

We propose that these patients should be differentiated from similar condition like Bloom syndrome, Rothmund-Thompson syndrome, xerodermapigmentosum, and progeria. monitor for cataract development and should be extracted early to prevent amblyopia.

Photophobia should be managed by providing tinted glasses. Patient with low vision should be managed with low vision aids.

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