

Cockayne Syndrome: A Rare Case Report.

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Abstract : The Cockayne Syndrome is a rare syndrome of congenital origin which is characterized by growth retardation, facial dysmorphism, facial naevi, retinopathy and mental retardation, which are associated with the changes in the brain parenchyma. The findings of CT/MRI of the brain support the clinical diagnosis of the Cockayne Syndrome. We report the CT findings of this rare syndrome.

INTRODUCTION

The Cockayne Syndrome which is transmitted as an autosomal recessive (DNA) disorder and it consists of abnormal ultra violet hyper sensitivity, retinitis pigmentosa, premature aging, dwarfism, microcephaly, deafness and progressive encephalopathy¹. The Cockayne Syndrome is a rare condition with less than 40 cases in the world literature^{1,3}!

CASE REPORT

A 3yr old male child born of 3rd degree consanguineous marriage with h/o developmental delay, red rashes over the face which increased on exposure to sunlight, frequent rubbing of face, one episode of febrile seizures at the age of 15 month with recurrent RTI presented to the dermatology department for rash. On examination, there is microcephaly, hair is sparse, brownish and lustreless, sunken eyes, beaked nose, hyperpigmented to erythematous plaques with scaling and multiple freckles over the malar region and bridge of nose, telangiectasias, chapping of lips exaggerated knee and ankle reflex. Opthexamn showed retinitis pigmentosa with delayed constriction of pupil on atropine. Serological tests are normal. ANA test was normal. Serological tests for TORCH infections was normal. CT brain revealed symmetric calcification involving B/L putamen, curvilinear cortical calcification at sulcal depths involving frontal, parietal, occipital lobes on both sides, parenchymal calcification, dentate nucleus calcification, diffuse cerebral/cerebellar atrophy, ex vacuo hydrocephalus



Fig :1. CT axial, coronal & sagittal view showing 1) Bilateral basal ganglia and parenchymal calcification 2) diffuse cerebral atrophy 3) ex vacuo hydrocephalus 4) Dentate nucleus calcification

DISCUSSION

The Cockayne Syndrome which is transmitted as an autosomal recessive (DNA) disorder. This syndrome often goes undetected in infancy, during which the growth and development are normal. The progressive degeneration is manifested by the second or the third year of life. Most of the survivors live up to the fourth decade. The male to female ratio is 3:1.

The most common neurological manifestation is mental retardation, and the other classical features include retinitis pigmentosa and photosensitive retinitis. The previously reported radiological findings of the Cockayne Syndrome include atrophy of the brain stem,

cerebellum and the cerebrum, and the high signal intensity of the cerebral white matter on T2 weighted images, which reflects demyelination². Calcification of the dentate nucleus of the cerebellum and the basal ganglia has also been documented.

The pathogenesis of the Cockayne Syndrome is unknown. Its diagnosis is based on the clinical diagnostic criteria and it may be supported by the demonstration of the intracranial calcification. The calcification of the basal ganglia and the subcortical white matter may be detected on CT as early as 3 years of age. The few detailed neuropathological studies which have been done, have shown calcification in the basal ganglia, with a variable degree of cerebral and cerebellar calcifications and cerebellar atrophy and white matter atrophy, with a patchy demyelination^{4,5}.

The CT in our patient showed diffuse white matter atrophy with no demyelination and hence made possible to differentiate the Cockayne Syndrome from the other white matter diseases in the absence of the intraparenchymal brain calcifications. In the appropriate clinical context, the MRI features like hypomyelination, supratentorial white matter loss, cerebellar atrophy or hypoplasia, and bilateral basal ganglia, dentate calcifications are an important adjunct for supporting the diagnosis of the Cockayne Syndrome.

Differential Diagnosis

(1) Fahr syndrome excluded patient because of early age; (2) mitochondrial/metabolic leucodystrophies; (3) Pseudohypoparathyroidism; (4) Blooms syndrome, Fanconis anemia, Xeroderma pigmentosa, Rothmund Thompson syndrome, Progeria; (5) SLE-ANA test

CONCLUSION

Hypomyelination, supratentorial white matter loss, cerebellar atrophy or hypoplasia, and bilateral basal ganglia calcifications are the most typical features in the Cockayne Syndrome, which are often associated with the cortical calcifications in the early-onset types of the disease. These features can help in differentiating the Cockayne Syndrome from other leukodystrophies⁵.

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Massive Pericardial Effusion in a 38 year old man secondary to Hashimoto's Thyroiditis: A Case Report.

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Abstract : Massive pericardial effusion (PE) was diagnosed in a 38 years old man which turned to be secondary to severe hypothyroidism. Pericardial effusion is well known in hypothyroidism, but it is rare as the sole presenting sign. After detailed history taking, physical examination, echocardiography and thyroid function survey, pericardial effusion secondary to Hashimoto's thyroiditis was impressed. Pericardiocentesis is unnecessary in most of the patients with hypothyroidism-associated pericardial effusion. It completely disappeared after 6-month thyroid hormone replacement. We recommend that hypothyroidism should be included in the differential diagnosis of unexplained pericardial effusion.

INTRODUCTION

The occurrence of pericardial effusion in hypothyroidism appears to be dependent on the severity of the disease. Pericardial effusion (PE) may be a frequent manifestation in myxedema (30-80%) but is rarely associated with mild hypothyroidism (1-6%)¹. Here we report a case of 38 year old male patient presenting with pericardial effusion as sole manifestation of severe hypothyroidism evaluated in our hospital ESIC-MC-PGIMSR.

CASE REPORT

A 38 year old male presented to outpatient department of our hospital with complaints of easy fatigability from last 6 months. On auscultation muffled heart sounds heard and on ECG low voltage complexes were present. Patient was subjected for 2D-echocardiography and chest radiograph. Depending on findings massive pericardial effusion was diagnosed and patient admitted for detailed evaluation.

On further questioning, he denied history of fever, chest pain, shortness of breath, paroxysmal nocturnal dyspnoea, jaundice, abdominal pain, haematemesis, urinary symptoms, fever, and anorexia or weight loss. He had no history of recent travel, or blood transfusion, or close contact with tuberculosis patient. There was no history of medical illness and drug history in the past.



Figure 1: Chest radiogram showed cardiomegaly with globular enlargement of the cardiac silhouette with "water bottle" configuration.

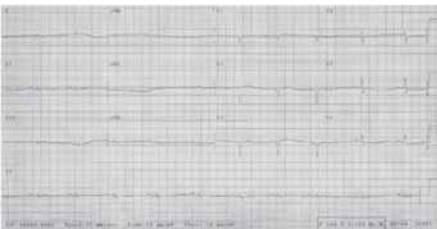


Figure 2: Electrocardiogram low voltage pattern.

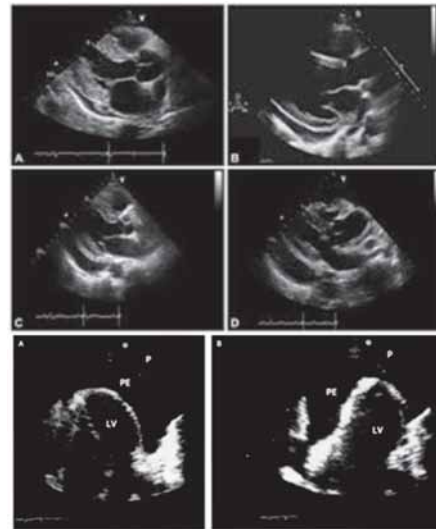


Figure 3: Echocardiogram demonstrated massive circumferential pericardial effusion with swinging motion of heart within large effusion.

On examination patient had dry skin, hoarseness of voice, apathy. There was no pedal edema. Vital signs on admission were recorded (temperature, 36°C; blood pressure, 110/70 mmHg, heart rate, regular at 64 beats/min). Body mass index (BMI) was 21.48kg/m². Oxygen saturation (SpO₂) was 98%. The cardiac apical impulse was not visible. The apex beat was not felt. The heart sounds were muffled. On neurological examination deep reflexes showed delayed relaxation. After the diagnosis of pericardial effusion was established, series surveys of etiologist were done. No malignancy, trauma or rheumatologic disorders was noted. Complete hemogram, liver and renal function and electrolyte were all within normal limits.

C-reactive protein was negative and anti-nuclear antibody revealed no significant findings.

Thyroid function test subsequently confirmed severe hypothyroidism- TSH 317 microIU/ml (normal range 0.28-6.82), T4 of 0.4(normal range 4.4-10.8µg/dl), T3 of 0.27(normal range 0.52-1.85µg/dl). Creatinine kinase and serum lactate dehydrogenase were normal.

Thyroid sonography showed diffuse coarse echogenicity over bilateral lobes suggestive of thyroiditis. Fine needle aspiration cytology showed thyroid follicular cells in clusters with background of pleomorphic population of lymphocytes multinucleate giant cells and fibrosis suggestive of hashimoto's thyroiditis. Anti thyroglobulin antibody was 25.22IU/ml (biological reference < 4.11 IU/ml) and anti-thyropoxidase antibody was 291.87 IU/ml (biochemical reference < 5.61 IU/ml). Thus, Hashimoto's thyroiditis was impressed. Lipid profile showed total cholesterol 268 mg/dl, triglyceride 243 mg/dl, HDL 48 mg/dl, LDL of 179mg/dl, VLDL of 49 mg/dl were noted. In consultation with endocrinologist he was started on low dose thyroxin which was increased gradually. Six months later, thyroid function and lipid profiles normalized gradually and follow-up echocardiography showed disappearance of previous pericardial effusion. Patient is doing well with OPD follow-up.

DISCUSSION

Pericardial effusion may be caused by acute pericarditis, tumor,

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