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Case Study

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COCKAYNE SYNDROME: NEED FOR MULTIDISCIPLINARY APPROACH

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ABSTRACT
A 7 year female child who was being managed as a case of Global developmental delay with spastic diplegia with seizure disorder, referred to dermatology for photosensitive rash on the cheeks since 6 years. Although the underlying abnormality appears to be autosomal recessive inheritance or metabolic (possibly thymic) dysfunction, there is no consensus on etiology. The multiple organ involvement carries significant implications for the anesthetist. Intubation can be technically difficult, and care of the skin can be problematic. Essential hypertension, hepatic deficiencies, osteoporosis, deafness, blindness, and other effects of premature aging may be encountered making perioperative management a challenge.

INTRODUCTION

Cockayne syndrome (also called Weber-Cockayne Syndrome or Neill-Dingwall Syndrome) is a rare autosomal recessive degenerative disease with cutaneous, ocular, neurologic and somatic abnormalities. The entity was first described in 1936 by Cockayne. Till now around 150 cases have been reported in the literature. The incidence in Western Europe has been evaluated as 2.7 per million births.[1,2]

Clinically different classes of CS have been distinguished: A classical form (CS I) which includes the majority of patients, a severe form (CS II) characterized by early onset and severe progression of manifestations and a mild form, typified by late onset and slow progression of disease. Classical CS patients show (a) growth failure, (b) neurodevelopmental delay and neurological dysfunction, (c) cutaneous photosensitivity, (d) progressive ocular abnormalities (pigmentary retinopathy, cataract), (e) hearing loss, (f) dental caries, (g) characteristic wizened facial appearance: bird-like facies. Intracranial calcifications are seen in some individuals. For diagnosis of CS in an infant, the presence of the first two criteria and a few of the other five criteria are required.[3]

CASE REPORT

- 7 year female child who was being managed as a case of Global developmental delay with spastic diplegia with seizure disorder, referred to dermatology for photosensitive rash on the cheeks since 6 years
- History of delayed developmental milestones
- History of inability to walk
- History of deviation of left eye since 6 years
- History of inability to gain weight
- History of pain and discharge from ears since 3 months
- Birth history was uneventful

GENERAL PHYSICAL EXAMINATION (Figure 1 to 3)

- Emaciated (decreased weight for age)
- Triangular facies
- Sunken eyes
- Decreased intercanthal distance
- Low set ears
- Retrognathia
- Microcephaly
- Flexion contracture both ankle joints)

MUCOCUTANEOUS EXAMINATION

- Faint erythema with brown colouerd macules over
- the cheeks giving mottled pigmentation pattern
- Poor condition of the dentition

CNS EXAMINATION

- Pupils reacting to the light
- Spasticity -both lower limbs
- Decreased muscle power
- Brisk deep tendon knee reflex
- Extensor plantar reflex
- Low IQ 43 (Imbecile)
- MRI brain-Paucity of white matter both cerebral hemispheres with atrophy of brainstem, corpus callosum and cerebellum

EYE EXAMINATION

- Alternating divergent squint 15 degree
- Pigmentary retinopathy

- No refractive error and cataract

ENT EXAMINATION

- Uncooperative for Pure tone audiometry

ECG-Normal

Figure 1&2: shows emaciated, triangular facies with sunken eyes, decreased intercanthal distance and low set ears.

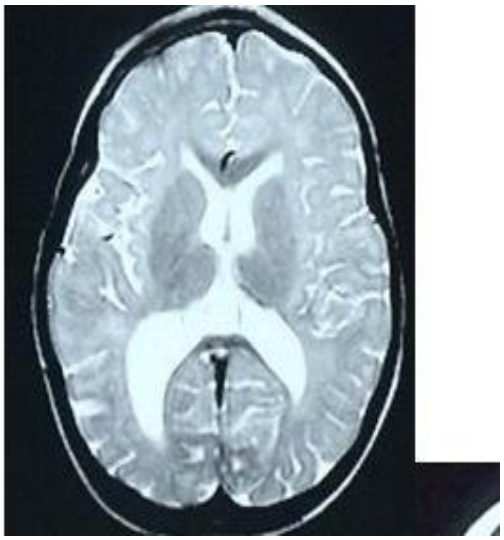

Figure 3: MRI brain showing paucity of white matter both cerebral hemispheres with atrophy of brainstem, corpus callosum and cerebellum.

DISCUSSION

The clinical features of Cockayne syndrome are discussed in the cases reported in the literature.[4] Importants neurological disorders, especially neuropsychiatric developmental delay, weakness, muscle atrophy, abnormal movements such as tremor and myoclonus, changes in tonus, seizures and headache have been described. Are characteristic of the syndrome: ocular changes as decreased lacrimation, cataracts, optic atrophy or hypoplasia. Other manifestations as hearing loss, nasal mucosa atrophy, hypertrophic tonsils and palate and dental abnormalities such as cavities, delayed tooth eruption, malocclusion, hypoplastic or absent teeth, are normally seen in this syndrome. The clinical evaluation and detailed physical examination are important aspects to suggest the diagnosis. It is also important to perform imaging tests such computed tomography or magnetic resonance image that can show or not changes in the white matter. Assessment should also be performed with an ophthalmologist to rule out ocular pathology associated with the syndrome as well as molecular genetic testing to confirm the Cockayne syndrome diagnosis.[5]

Specific tests for the syndrome were not performed in this case report, but the suggestive clinical picture and two imaging exams compatibles with the syndrome were key figures that suggested the diagnosis. The importance of addressing the issue described is to consider the main clinical characteristics of the disease in order to think in this diagnosis in potential. In this situation, it is desirable a limited sun exposure, physical therapy to reduce

muscular symptoms, besides evaluation and genetic counseling of the patient's family.[6]

CONCLUSION

As CS is a multisystem DNA repair disorder so we require sytemic approach to diagnose the disease at an early stage as patient was being treated on referral basis only.

DECLARATION OF PATIENT CONSENT

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/ their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

CONFLICT OF INTEREST

Nil.

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