PACIFIC JOURNAL OF MEDICAL SCIENCES

{Formerly: Medical Sciences Bulletin}

ISSN: 2072 – 1625

Pac. J. Med. Sci. (PJMS)

www.pacjmedsci.com. Email: pacjmedsci@gmail.com.

COCKAYNE'S SYNDROME: A CASE REPORT

***RAGHAVENDRA M N, **ANURADHA PAI, ***SURESH K V, ****YUSUF AHAMMED. A. RONAD, *****MOUNESH KUMAR C D**

*Department of Oral Medicine & Radiology, Vishnu Dental College, Bhimavarum Andra Pradesh, India;
**Department of Oral Medicine & Radiology, Oxford Dental College and Hospital, Bangalore, Karnataka, India;
***Department of Oral Medicine & Radiology,
School of Dental Sciences, Krishna Institute of Medical Sciences, Deemed University, Karad, Satara (District), Maharashtra (State), India;
****Department of Orthodontics & Dentofacial Orthopaedics, School of Dental Sciences, Krishna Institute of Medical Sciences, Deemed University, Karad, Satara (District), Maharashtra (State), India;
*****Department of Oral and maxillofacial surgery, School of Dental Sciences, Krishna Institute of Medical Sciences, Deemed University, Karad, Satara (District), Maharashtra (State), India.

Correspondence author: Dr K. V. Suresh: dr_suri88@yahoo.co.in. dr.suri88@gmail.com.

Running Title: Raghavendra et al. Cockayne's syndrome

COCKAYNE'S SYNDROME: A CASE REPORT

***RAGHAVENDRA M N, **ANURADHA PAI, ***SURESH K V, ****YUSUF AHAMMED. A. RONAD, *****MOUNESH KUMAR C D**

*Department of Oral Medicine & Radiology, Vishnu Dental College, Bhimavarum Andra Pradesh, India;
**Department of Oral Medicine & Radiology, Oxford Dental College and Hospital, Bangalore, Karnataka, India;
***Department of Oral Medicine & Radiology,
School of Dental Sciences, Krishna Institute of Medical Sciences, Deemed University, Karad, Satara (District), Maharashtra (State), India;
****Department of Orthodontics & Dentofacial Orthopaedics, School of Dental Sciences, Krishna Institute of Medical Sciences, Deemed University, Karad, Satara (District), Maharashtra (State), India;
*****Department of Oral and maxillofacial surgery, School of Dental Sciences, Krishna Institute of Medical Sciences, Deemed University, Karad, Satara (District), Maharashtra (State), India.

Correspondence author: Dr K. V. Suresh: dr_suri88@yahoo.co.in. dr.suri88@gmail.com.

Running Title: Raghavendra et al. Cockayne's syndrome

ABSTRACT:

Cockayne's syndrome (CS) is a rare, autosomal recessive disease resembling progeria. The features of CS do not appear until 4 to 5 years of age. Most patient presents with cachectic dwarfism, cutaneous photosensitivity, loss of adipose tissue, mental retardation, skeletal and neurological abnormalities, similar to the current case. The additional feature observed in the present case was actinic chelitis. We report a case of Cockayne's syndrome with pronounced oral manifestations and an unusual feature of actinic chelitis.

Key words: Cockayne Syndrome, Actinic Cheilitis, Genetic Disorder, Progeria

(Submitted April 2012; Accepted June 2012)

INTRODUCTION:

Cockayne's syndrome (CS) is a rare autosomal recessive disorder described first by Cockayne in 1936 [1]. It has been proposed that mutation of two genes, CNK1 (ERCC8) and ERCC6, located on the 5 and 10 chromosomes respectively may lead to two variations of CS [1]. The major features of CS include progressive loss of muscle and subcutaneous

tissue, short stature, premature aging, senile face, mental retardation, microcephaly, retinopathy, beak-like nose, hearing loss, carious teeth, relatively large hands and feet, joint contractures, photosensitive dry skin, and thin hair [2]. Progression of CS leads to functional debility and bed ridden in the second decade of life. Patient eventually dies because of respiratory and other infections [3]. Patients affected by this syndrome usually have unfavorable prognosis. Early diagnosis is most crucial, for proper genetic counseling and antenatal screening [4]. The present case is relevant because it is a rare condition and presents important oral findings.

CASE REPORT:

A 14 year old male patient, born of a consanguinous marriage, was brought to the hospital by his father for complaint of wound on the lower lip since two years. The wound was associated with provoked bleeding and moderate pain. Medical history revealed that, before visiting our department the patient was hospitalized for 10 days, as he was suffering from tremors and difficulty in speech. Patient's father reported that the child was given treatment in the local hospital for chronic suppurative otitis media at the age of 5 years and 10 years. Neonatal history revealed lower segment cesarean term, birth weight 2.75kg and no post natal complications. Anthropometry revealed weight 15.5kg and height 119.0cm. Ataxia, cerebellar signs, gait abnormality, horse riding stance were present (Fig. 1). Magnetic resonance imaging of the head and brain showed mild cerebrum and cerebellum atrophy (Fig. 2); no abnormality was detected with chest and hand radiographs. Blood cell count and serological values were within normal limits. Ophthalmic evaluation revealed evidence of exposure keratitis started lacryge. On examination, an excoriated lesion with vertical fissuring and crustations were apparent on vermillion zone of lower lip and labial mucosa (Fig. 3). Gingiva was soft and edematous, partial macrodontia principally of central incisors, generalized dental fluorosis with hypoplasia and deep palate were noticed. Panoramic revealed bilateral condylar hypoplasia. Cytogenic reference was opined as cockayne syndrome. Actinic cheilitis was set as the provisional diagnosis for the lip lesion and patient was advised to apply sunscreen cream with sun protection factor 15. After a week the patient was reexamined, there was marked regression of the lesion. Patient was advised to avoid unnecessary exposure to the sunlight and to go for regular health check up. Patient informed consent and Ethical committee clearance was obtained for the present case report.

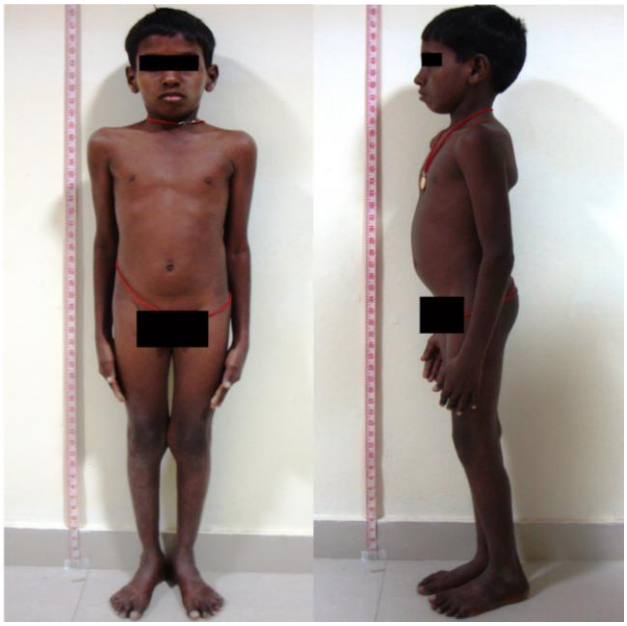

Fig 1: Height, gait and horse riding stance

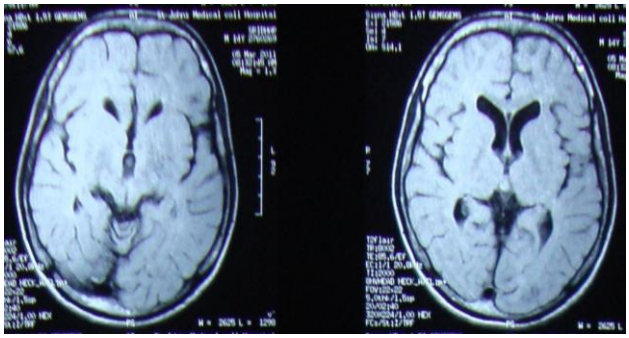

Fig 2: MRI images of brain showing mild cerebrum and cerebellum atrophy

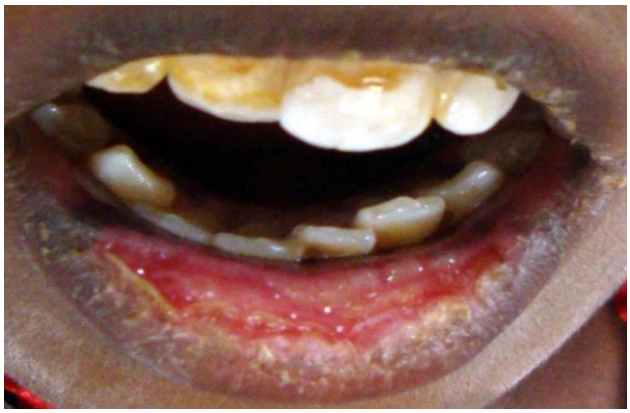

Fig 3: Photograph showing excoriated lesion in Vermillion zone of the lower lip with vertical fissuring and crustations.

DISCUSSION:

Xeroderma pigmentosum (XP), Cockayne syndrome (CS) and trichothiodystrophy (TTD) are genetic disorders with very different clinical features, but they are associated with defects in nucleotide excision repair [5]. For affected individuals at age of 2 to 4 years, CS usually becomes evident with changes in personality and behavior [4]. Mental deterioration is progressive and the dwarfism becomes obvious at this time. A characteristic facies develops, resulting in a thin prominent nose, prognathism, sunken eyes, and a lack of subcutaneous fat. Other major neurological abnormalities include sensorineural hearing loss, ataxia, spasticity, myoclonus, and gait disturbance [6]; similar characteristic findings were noticed in the present case.

The usual oral findings are delayed deciduous teeth eruption, oligodontia, short roots, more incidence of caries, a deep palate, atrophy of the alveolar processes, mandibular prognathism and condylar hypoplasia [7]. We noticed macrodontia of central incisors, generalized dental hypoplasia and condylar hypoplasia in the present case.

The diagnosis was basically clinical, though supportive diagnostic tests were available including computerized tomography brain. The prognosis for affected patients is poor, with high mortality rate; most affected children may die by the second decade of life, if appropriate supportive measures are provided [8].

CS patient frequently develops photosensitivity dermatitis that results in desquamation and scarring of the skin. Our patient suffered from actinic chelitis of lower lip, which may be because of excessive exposure to the sunlight. A prematurely aged metabolic state has also been hypothesized as one of the principal causes of CS [9].

The CS can be diagnosed prenatally by examining amniotic cells cultured in vitro. The prenatal test can be carried out two to four days after the culture of sufficient cells [10].

CONCLUSION:

Anomalies of craniofacial and oral region are common in CS, and also life expectancy for the individuals suffering from this syndrome is comparatively short. Dentist should play a key role in the early diagnosis and management of oro-dental anomalies. Frequent examinations and emphasis on preventing dental disease must be stressed to the parents because of the difficulty in providing restorative care. Appropriate and safe dental care for patients

with Cockayne's syndrome can be rendered after medical consultation.

REFRENCES:

1. María de la Luz Arenas Sordo, Edgar Hernández Zamora, Luis Alberto Montoya Pérez, Beatriz Catalina Aldape Barrios. Cockayne's Syndrome: A case report. Literature review. Oral Medicine and Pathology E236-238.
2. Adel A.H. Mahmoud, George M. Yousef, Ibrahim Al-Hifzi, and Eleftherios P. Diamandis. Cockayne Syndrome in Three Sisters with Varying Clinical Presentation. American Journal of Medical Genetics 2002; 111:81–85.
3. M. Mohammadi cockayne syndrome: report of two cases within a family. Acta medica iranlca, 1999; 37 (2), 115-
4. Lehmann AR. DNA repair-deficient diseases, xeroderma pigmentosum, Cockayne syndrome and trichothiodystrophy. Biochimie 2003; 85:1101-11.
5. Tuo J, Chen C, Zeng X, Christiansen M, Bohr VA. Functional crosstalk between hOgg1 and the helicase domain of Cockayne syndrome group B protein. DNA Repair (Amst). 2002; 1:913-27.
6. Robert A. Boraz, Cockayne's syndrome: literature review and case report. Pediatric Dentistry: July/August, 1991 N Volume" L 3, No 4; 227-230.
7. Jones KL: Smith's Recognizable Patterns of Human Malformation. 4th ed. Philadelphia: WB Saunders Co, 1988, pp 122-23.
8. Kennedy RM, Rowe VD, Kepes JJ: Cockayne syndrome, an atypical case. Neurology 30:1268-72, 1980.
9. Ohno T, Hirooka M: Renal lesions in Cockayne's syndrome. Tohoku J Exp Med 89:151-66, 1966.
10. Lehman AR, Francis AJ, Giannelli F: Prenatal diagnosis of Cockayne's syndrome. Lancet 1:486-88, 1985.