IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 17, Issue 11 Ver. 8 (November. 2018), PP 43-45
www.iosrjournals.org

Case report on a rare case of Cockayne Syndrome

Dr.Prerna Hasani[1*], DrRishabh Bhatia [2]*, Dr.N.A.Patel[3*]
[1]Third year Resident, [2]* Third year Resident, [3*]Assistant Professor, Department of Radiology, B.J.Medical College, Civil Hospital, Ahmedabad, Gujarat, India.*
Corresponding Author: Dr.Prerna Hasani

Abstract: *Cockayne syndrome is a rare autosomal recessive, neurodegenerative disorder characterized by symptoms like microcephaly, failure to thrive, photosensitivity, hearing loss, vision loss, severe tooth decay and bony abnormalities. The brain imaging findings are cardinal features, which helps to arrive at the diagnosis of this rare disorder. Calcifications and brain atrophy were the main imaging features on noncontrast CT scans. Calcifications were typically found in the basal ganglia and less often in the cortex and dentate nuclei. Here in this article, we would like to highlight the NCCT Brain findings of Cockayne syndrome.*

Key Words: *Cockayne syndrome, Developmental delay, Calcification, Atrophy*

Date of Submission: 14-11-2018 Date of acceptance: 29-11-2018

I. Case Report

A 5 year old female child presented with the complaint of gross developmental delay. On physical examination, the patient had microcephaly, short stature and global developmental delay. On eliciting family history, younger sibling of the patient, 3 year old male child, has similar complaints. Audiometric assessment revealed bilateral sensoryneural hearing loss. Fundoscopic examination revealed bilateral disc pallor and pigmentory changes suggestive of atypical retinitis pigmentosa. The patient was then subjected to NCCT Brain study. NCCT Brain was done under sedation, which revealed bilateral symmetrical hyperdensities involving lentiform nuclei, lateral part of thalami, dentate nuclei and subcortical white matter of fronto-parieto-occipital regions suggestive of soft calcifications. Prominent sulci, fissures, basal cisterns and cerebellar folia are seen, with mild dilatation of all four ventricles, indicative of cerebral cortical and cerebellar atrophy.

The clinical and NCCT Brain features helped in arriving at the diagnosis of cockayne syndrome.

II. Discussion

Cockayne Syndrome is a rare multisystem disorder, with autosomal recessive inheritance. It belongs to the family of nucleotide excision repair diseases. Cockayne syndrome can result from mutations in either the ERCC6 gene (also known as CSB) or the ERCC8 gene (also known as CSA). These genes provide instructions for making proteins that are involved in repairing damaged DNA.[1]

Clinical criteria required for the diagnosis includes poor growth and neurologic abnormality; other very common manifestations include sensorineural hearing loss, cataracts, pigmentory retinopathy, cutaneous photosensitivity, and dental caries.[2]

Neuroimaging features of Cockayne Syndrome:

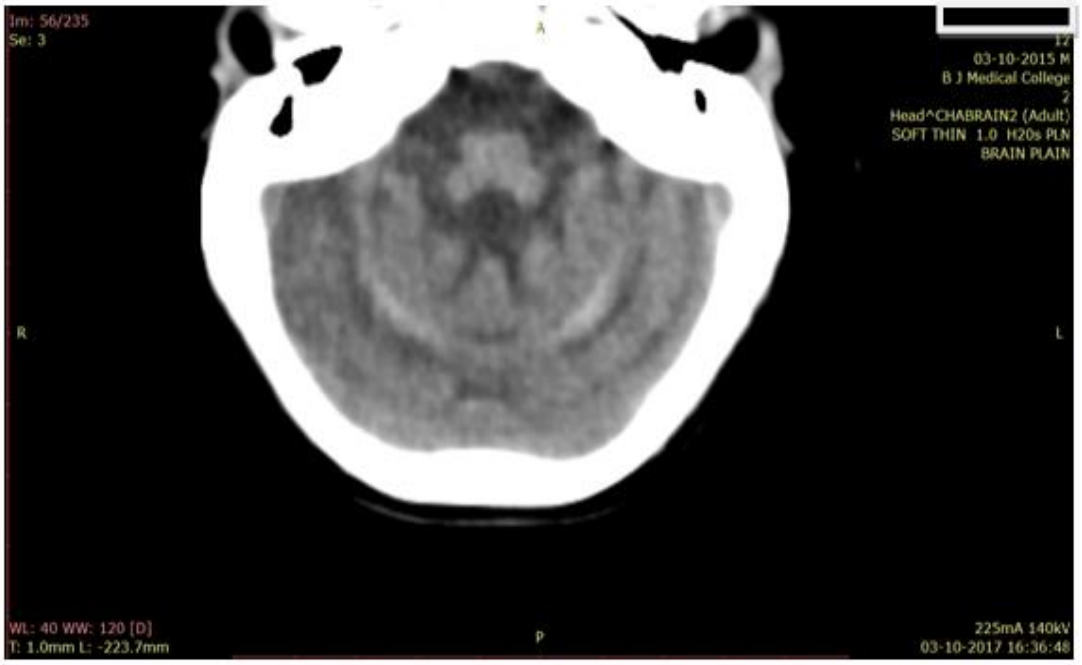
The previously reported radiological findings of the Cockayne Syndrome include atrophy of the brain stem, cerebellum and the cerebral parenchyma. [3-7] Calcification of the dentate nucleus of the cerebellum and the basal ganglia has also been documented. [5-7]

Diagnosis of the cockayne syndrome 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 scan 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 cerebral as well as cerebellar atrophy. [8]

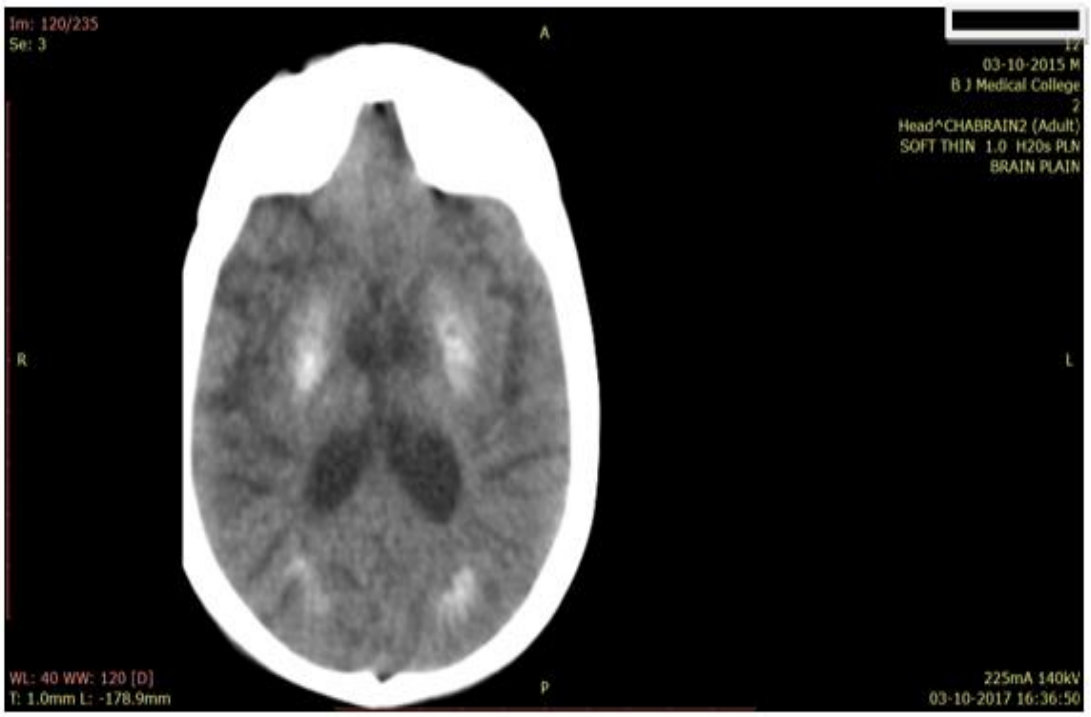
The NCCT Brain study in our patient showed calcification in bilateral thalami, lentiform nuclei, dentate nucle and subcortical white matter in bilateral fronto-parieto-occipital regions associated with cerebral cortical as well as cerebellar atrophy.In the appropriate clinical context, the imaging features like cerebellar atrophy, bilateral basal ganglia calcifications are an important adjunct for supporting the diagnosis of the Cockayne Syndrome.

III. Conclusion

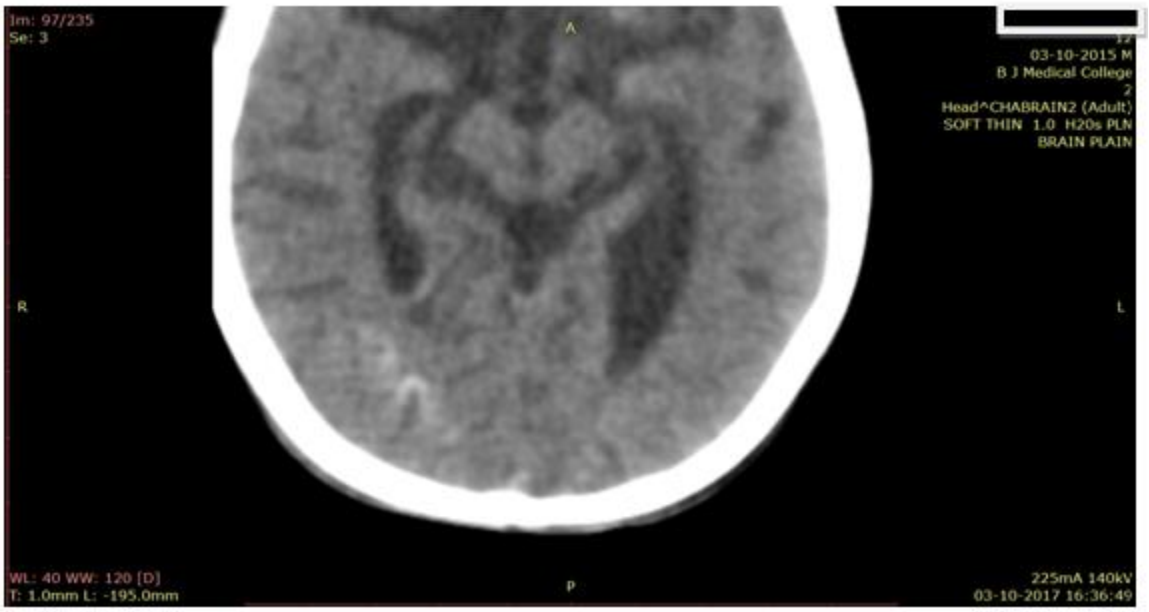
Cerebellar atrophy and bilateral basal ganglia calcifications are the most typical features in the Cockayne Syndrome, which are often associated with the cortical calcifications in the early-onset types of the disease. These features can help in differentiating the Cockayne Syndrome from other causes of leukodystrophies.



Calcification in dentate nucleus of cerebellum.



Calcification in putamen and cerebral region mainly in subcortical region. Note the symmetry of calcification.



Typical subcortical calcification.

References

[1]. Nouspikel T[1]. Nucleotide excision repair and neurological diseases.DNA Repair (Amst). 2008 Jul 1;7(7):1155-67. doi: 10.1016/j.dnarep.2008.03.015. Epub 2008 May 5.
[2]. M.D. Martha A. Nance, Susan A. Berry.Cockayne syndrome: review of 140 cases. *A J Med Genet* 1992*;* 42*:* 68– 84
[3]. Sugita K, Takanashi J, Ishii M, Niimi H. Comparison of the MRI white matter changes with the neuro psychological impairment in the Cockayne Syndrome. Paediatr Neurol. 1992;8:295–98.
[4]. Hamel BC, Raama A, Schuitema-Dijkstra AR, et al. The Xerodermapigmentosa – the Cockayne Syndrome complex: a further case. J Med Genet. 1996;33:607–10.
[5]. Demaerel P, Kendall BE, Kingsley D. Cranial CT and MRI in the diseases with DNA repair defects. Neuroradiology. 1992;34:117–21.
[6]. Bolthauser E, Yalcinakya C, Wichmann W, Reuuter F, Prader A, Valavanis A. MRI in the Cockayne Syndrome Type 1. Neuroradiology. 1989;31:276–77.
[7]. Demaerel P, Wilms G, Verdru P, Carton H, Baert AL. MRI in the diagnoisis of the Cockayne Syndrome. One case. J Neuroradiol. 1990;17:157–60.
[8]. Softer D, Grotsky W, RapinI, Suzuki K. The Cockayne Syndrome: the unusual neuropathological findings and the review of the literature. Ann Neurol. 1979;6:340–48.

Dr.Prerna Hasani. "Case report on a rare case of Cockayne Syndrome" IOSR Journal of Dental and Medical Sciences (IOSR-JDMS), vol. 17, no. 11, 2018, pp 43-45.