

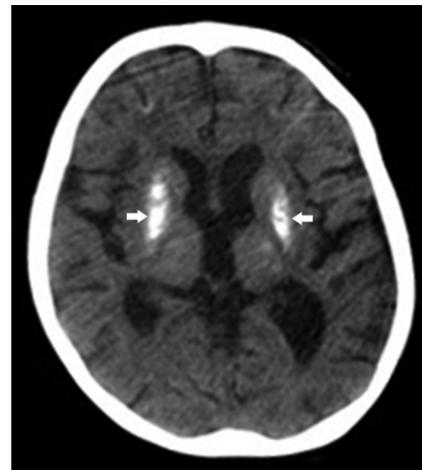
# Cockayne syndrome with intracranial calcification, hypomyelination, and cerebral atrophy

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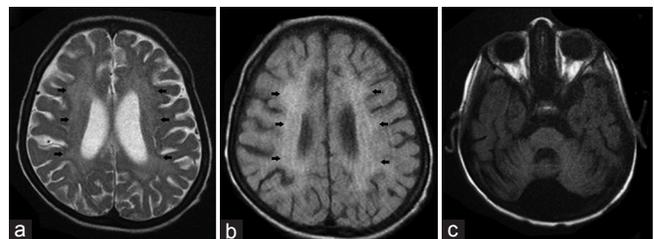
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A 5-year-old boy with global developmental delay was admitted with ataxia of 2-month duration. On examination, he had microcephaly and dysmorphic facies with large ears and sunken eyes. CT head revealed bilateral basal ganglia calcification [Figure 1]. Magnetic resonance imaging brain showed atrophy of cerebrum and hypoplasia of cerebellum, with bilateral symmetrical T2 and T2-fluid-attenuated inversion recovery hyperintensities in periventricular white matter suggestive of hypomyelination [Figure 2]. A diagnosis of Cockayne syndrome (CS) was made based on clinico-radiological features.

CS is a rare multisystem disorder, with autosomal recessive inheritance. It is caused by mutation in either or the two genes *CSA* or *CSB*, which is involved in nucleotide excision repair pathway of DNA repair mechanism. Features of CS include physical and mental growth retardation due to postnatal arrest in brain development, characteristic facies with progressive loss of fat and deep set eyes, ocular abnormalities such as cataracts, pigmentary retinopathy and optic atrophy, sensorineural hearing loss, and cutaneous hypersensitivity to ultraviolet rays, but without increased risk of cancer.<sup>[1]</sup> Imaging findings of CS include bilaterally symmetric calcifications, especially in the putamen, hypomyelination of the supratentorial white matter most pronounced in the periventricular area,



**Figure 1:** Computed tomography head showing bilateral basal ganglia calcification (arrows)



**Figure 2:** Magnetic resonance imaging brain - axial T2-weighted image (a) and fluid-attenuated inversion recovery imaging (b) showing periventricular hyperintensity suggestive of hypomyelination (arrows). Cerebellar hypoplasia is also evident in the images. Axial T1-weighted image (c) at the level of pons showing hypoplasia of cerebellum

global cerebral atrophy with ventricular dilatation and cerebellar atrophy.<sup>[2]</sup>

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### Conflicts of interest

There are no conflicts of interest.

### References

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