Melanin, but also slow nail growth rate resulting in larger accumulation of the pigment. The presence of HHN neither correlates with degree of renal impairment nor with blood urea nitrogen or creatinine levels. HHN can be an important clue in making the diagnosis of renal disease.

References

Schizencephaly

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Fig. 1: Optic atrophy in Rt eye
Fig. 2: Axial T2W MR image at the level of body of lateral ventricles demonstrates absent septum pellucidum
Fig. 3: Coronal T2W MR image demonstrating a CSF filled cleft lined by gray matter in the right inferior frontal gyrus, not communicating with the ipsilateral ventricular system

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30 year old male presented with history of generalised tonic clonic seizures since the age of 16 poorly controlled with antiepileptic drugs and involuntary eye movements observed by his parents since childhood. He is the third child born at full term to a non-consanguineous couple with no family history of genetic disease and mental retardation. No history of delayed milestones. There was no history of fever, radiation exposure or any significant drug intake or any major illness during pregnancy period of his mother. On examination patient was conscious and co-operative. Vitals were normal. Neurological examination revealed normal higher mental functions, Mini mental score of 29. Visual acuity Rt eye was 6/60, Lt eye was finger counting at 1 meter. Fundus examination revealed optic atrophy in both eyes (Figure 1). Horizontal nystagmus present in both eyes on looking at both directions. Ocular movements were normal, pupils were bilaterally equally reacting normally. Rest of the CNS examination was normal. Respiratory, cardiovascular and abdominal examination was unremarkable. Male external genitilia and secondary sexual characteristics were normal. Routine investigations like haemogram, blood sugar, LFT, RFT, Electrolytes, ECG were normal. MRI brain with orbits showed absence of septum pellucidum, CSF filled cleft lined by gray matter noted in the right inferior frontal gyrus, not communicating with the ipsilateral ventricular system suggestive
of closed lip schizencephaly (Figures 2 and 3). Both optic nerves are small in size and optic chiasma is small in size. Rest of the brain parenchyma reveals normal signal characteristics. Ventricular systems show no dilatation or distortion. The cervicomedullary junction is normal with no tonsillar herniation. EEG was normal.

Schizencephaly is a rare developmental disorder of neuronal migration in which there are clefts spanning the cerebral hemisphere which are characterised by an infolding of the grey matter along the cleft from the cortex into the ventricles. The incidence is 1.5 in 100,000 live births. There is no gender predilection. The lesion is most likely related to multiple aetiologies including genetic, toxic, metabolic, vascular or infectious agents. The clefts may be unilateral or bilateral. There are 2 types: closed lip (Type I) in which the walls of the cleft are fused, open lip (Type II) in which the walls of the cleft are separated. It is associated with defective expression of the gene EMX2. In closed lip (unilateral case) there is mild hemiparesis and seizure but normal development. In open lip, there is mild to moderate developmental delay with hemiparesis. In bilateral clefts there is severe mental deficits, severe motor anomalies including spastic quadriparesis. Frequently these patients present with blindness often associated with optic nerve hypoplasia. Most of the cases (80-90%) present with absent septum pellucidum. Treatment consists of control of seizures, physiotherapy, and in cases that are complicated by hydrocephalus, a ventriculoperitoneal shunt is needed.

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References