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Rud's Syndrome: A case report

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RUD's syndrome is a rare hereditary disorder of predominantly autosomal recessive inheritance, characterised by short stature, hypogonadism, ichthyosis, epilepsy and low intelligence quotient (IQ). A 14 year old adolescent male born to consanguineous parents presented with infantile genitalia, history of seizures since childhood and was taking antiepileptics (Carbamazepine 30mg/kg/day). He also had a very dry scaly ichthyosiform skin (Fig 1) since birth, which aggravated during winter and was put on emollients. His scholastic performance was very poor. On general examination, he was short statured with height being below the 3rd centile as per the WHO growth charts and weight being 23 kgs. He had small sized genitalia (Fig 2) with no development of secondary sexual characteristics. However, systemic examination was unremarkable. On thorough investigations, complete blood picture was normal, ophthalmic and hearing assessment revealed no abnormality. Ultrasonography of the abdomen was normal. Hormonal assay was abnormal with testosterone (0.025ng/mL), FSH (0.08mIU/mL) and LH (<30mIU/mL) being extremely low suggesting hypogonadotropic hypogonadism whereas thyroid function tests were normal. EEG was abnormal with bilateral spikes and waves. On developmental assessment, it was observed that the child had a low to borderline IQ (50-60). Based on the history, physical examination and laboratory investigation the diagnosis of RUD's syndrome was established and the child was being treated symptomatically and was advised to follow up periodically as there is no specific treatment for this disorder and multidisciplinary approach remains the mainstay of management.

**Fig 1.** Severe ichthyosis**Fig 2** Infantile genitalia

Biography

Nayantara Ro Gandra obtained bachelor degree in Medicine & Surgery (MBBS) from the NTR university of Health sciences, Andhra Pradesh, India in the year 2007. She did her MD (Pediatrics) from the same university in 2013. Currently, she working as an Assistant professor(Pediatrics) since 4 years at Apollo Institute of Medical Sciences and Research, Hyderabad, India. She has 8 publications to her credit in international and national journals.

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