Dear Editor,

Ring chromosome 20 syndrome (R20 syndrome) is a rare chromosomal disorder linked with epilepsy, described as early as 1972 and around 150 such cases have been reported in the existing literature.[1,2] R20 syndrome is characterized by childhood-onset intractable epilepsy, dysmorphic features, mental disability, behavioural issues and repeated non-convulsive status epilepticus (NCSE).[3,4] A cytogenetic test is required to confirm the diagnosis.

We evaluated a 21-year-old right-handed girl for drug-resistant epilepsy (DRE). Her events started at the age of 10 years. She had recurrent episodes of prolonged behavioural arrest lasting for 30–40 min during which she will be staring in unresponsive state throughout the event. The frequency of these events was 1–3 per day occurring during both awake and sleep. Her prenatal, birth, development and past medical history were unremarkable. She has no family history of epilepsy. Her scholastic performance was poor after the onset of epilepsy. On neuropsychological evaluation, her intelligence quotient (IQ) was 72, indicating borderline intelligence. No behavioural problems were reported. Her general and neurological examination was unremarkable. The magnetic resonance imaging (MRI) was normal and fluorodeoxyglucose-positron emission tomography (FDG-PET) was unremarkable.

She underwent long-term video electroencephalogram (EEG) to characterize the events. Many events were recorded. The seizure semiology was characterized by sudden onset of behavioral arrest with prolonged staring and immobile upper and lower extremities for 30–40 min associated with bilateral eye blinking. She was unresponsive to response testing throughout the event. Interictal EEG recording showed bi-frontal spike and sharp wave discharges [Figure 1a]. Ictal EEG recordings showed bi-frontal slow spike and wave discharges of 3–4 Hz frequency for 1–2 min followed by 5–6 Hz bifrontal spike and wave discharges which consistently evolved in frequency and amplitude for the next 30–40 min without any clear cut offset. The total duration of paroxysms was >40% of the recording length [Figure 1b].

A possibility of R20 syndrome was considered based on the above findings. To confirm the diagnosis, a chromosomal analysis of 30 lymphocytes was completed, which revealed a female karyotype with a translocation involving chromosome 1 and chromosome 12 along with a ring chromosome 20 in the mosaic condition in 14 of the 25 metaphases (56%) t(1;12)(q21;q11), mos r(20) [Figure 2]. Translocation involving chromosome 1 and 12 had not been linked to Epilepsy.

Previous treatment with valproic acid, oxcarbazepine and clobazam had no beneficial effect in reducing her seizure frequency. She was started on Valproic acid and Lamotrigine. At one year follow up, she had a significant reduction in seizure frequency with a frequency of 1–2 episodes per month.

R20 syndrome is characterized by distinct electro-clinical features such as (a) childhood-onset medically refractory...
epilepsy (b) mental disability (c) behavioral issues (d) dysmorphic features (e) focal seizures associated with oro-alimentary automatisms, frightened expression, altered consciousness with a staring gaze and frontal onset discharges of short periods (f) normal or near-normal background EEG activity with intermittent trains of constant theta waves in the fronto-central regions which may or may not be affected by eye-opening, level of consciousness or intravenous injection of diazepam (g) multiple episodes of NCSE with a classical EEG pattern characterized by serial runs of long-lasting unilateral or bilateral paroxysmal high-voltage slow waves with or without intermittent spikes over the frontal lobes which may spread over the scalp during the ictal event and (h) a normal brain MRI in most cases.\[5,6\]

Ring chromosomes have been identified for each of the 26 human chromosomes but only the ring chromosome syndromes that involve chromosomes 14, 17 and 20 are linked to epilepsy.\[3\] In R20 syndrome, epilepsy has an age-dependent course and has worse results when the initial age at seizure onset is earlier.\[2\] The EEG showing paroxysmal bursts during minutes with a total duration of paroxysms >20% of

Figure 1: The inter ictal EEG (a) showing bifrontal spike and sharp wave discharges The ictal EEG (b) showing runs of high-voltage bifrontal slow spike and wave activity (low frequency: 1 Hz; high frequency: 70 Hz; sensitivity: 7 µV/mm; paper speed: 30 mm/s)

Figure 2: A female karyotype with a translocation involving chromosome 1 and 12 (arrows) and a ring chromosome 20 (arrow) is seen
Acute ataxia and myoclonus in COVID-19: A case series

In 2019 (COVID-19) has been rarely reported. With other viral infections, myoclonus in coronavirus disease frequently published in literature. Amongst the various neurological manifestations, acute syndrome coronavirus 2 (SARS-CoV-2) have been reported. Neurological complications of the severe acute respiratory inflammation. Causes including postinfectious cerebellar immune-mediated. Acute ataxia and myoclonus in adults have a variety of semiology and NCSE can lead to delay in diagnosis. The evolving electroclinical syndrome of “epilepsy with ring chromosome 20”. Seizure 2012;21:92-7. Kalane U, Datar C, Kalane S. Ring chromosome 20 syndrome-A rare chromosomal cause of refractory epilepsy in children. Int J Epilepsy 2017;4:87-9. Chawla J, Sucholeiki R, Jones C, Silver K. Intractable epilepsy with ring chromosome 20 syndrome treated with vagal nerve stimulation: Case report and review of the literature. J Child Neurol 2002;17:778-80.

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Conflicts of interest
There are no conflicts of interest.

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