

## Ring 20 syndrome- A rare case report

Sumit Bhateja<sup>1\*</sup>, Geetika Arora<sup>2</sup>

Reader, <sup>1</sup>Dept. of Oral Medicine and Radiology, Manav Rachna Dental College, Faridabad, Haryana, <sup>2</sup>Reader, Dept. of Public Health Dentistry, Inderprastha Dental College & Hospital, Sahibabad, Uttar Pradesh, India

**\*Corresponding Author:**

Email: bhateja.sumit@gmail.com

### Abstract

The Clinical, electroencephalographic, neuroimaging (brain magnetic resonance image, MRI) findings of a young male patient with a rare cytogenetic anomaly known as Ring 20 chromosome syndrome characterized by mental retardation, seizures, emotional lability, without any other significant dysmorphies is documented.

**Keywords:** Ring 20 chromosome, EEG, Seizures.

### Introduction

Every cell has 46 chromosomes, 23 inherited from maternal and 23 from paternal. These are further also grouped as 44 autosomes and a pair of allosomes (sex chromosomes).<sup>1</sup> Each chromosome contains genes. Chromosomal syndromes can be broadly grouped as follows:

1. Duplication syndromes, with an additional segment of chromosome material.
2. Deletion syndromes, where a segment is lost.
3. Breakpoint disruption syndromes, where only few genes may be mutated.

Ring 20 syndrome is a rare genetic condition caused by an abnormal chromosome 20 that forms a ring. Every cell has a pair of chromosome 20. Ring 20 syndrome presents with “**mosaicism**” where some cells are affected and others are not.<sup>2</sup>

### Case Report

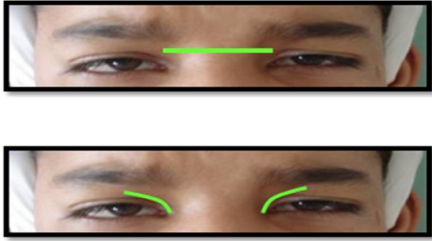
A 12 year old boy reported with a chief complaint of decayed tooth in right lower back tooth region since one year (Fig. 1). Anamnesis revealed patient is epileptic and prone to develop epileptic attacks by mental stressors. The first seizure was detected at around 18 months of age. He was born to parents when father's age was 26 year old and mother's age was 23 years. The couple is healthy and non-consanguineous, and the pregnancy was apparently normal. The labour was by caesarian section due to wrapped umbilical cord. The weight at birth was 2.6 kg. He was the first birth of an offspring of two. There was no history of similar case in the family reported.

Patient presented with delayed psychomotor development with hypotonia and stunted growth. Extraoral examination revealed coarse facial features, hypertelorism, slanting of eyes, plagiocephaly, sialorrhoea (Fig. 1, 2, 3, 4). Intraoral soft tissue examination did not reveal any significant findings except a high arched palate (Fig. 5). Intraoral hard tissue examination revealed missing 15, 17, 27. Root remnant i.r.t 85 with moderate stains and calculus was

also reported. An O.P.G investigation was advised for complete screening of the jaws. Patient was referred to Neurology Department of Medical College, Agra, India. Patient was advised for an MRI (brain) (Fig. 6a & 6b) and EEG (Fig. 7). MRI findings revealed that Corpus callosum showed focal thinning in the region of the junction of posterior third of the body with splenium. Electroencephalogram (EEG) findings revealed Delta and theta waves in anterior regions at times accompanied by spikes in temporal areas with variable lateralization on a generally slow basal activity which were suggestive of primary generalized epilepsy with possible myoclonal component. Based on history, clinical findings and Neurologist's consultation patient was diagnosed with Ring 20 chromosome syndrome. The dental management plan of patient included extraction of root remnant with respect to 85 and Oral prophylaxis. For the management of sialorrhoea-Oral motor training, appliance therapy and Scopolamine transdermal patches were advocated.



**Fig. 1: Coarse facial features**



**Fig. 2: Hypertelorism and slanting of eyes respectively**



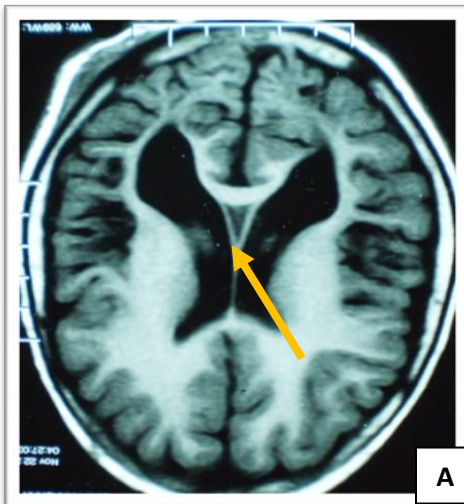
**Fig. 4: Sialorrhoea**



**Fig. 3: Plagiocephaly**



**Fig. 5: High arched palate**



**Fig 6 (A & B): MRI Brain: Corpus callosum shows focal thinning in the region of the junction of posterior third of the body with splenium**



**Fig. 7: EEG Findings which are suggestive of primary generalised epilepsy with possible myoclonal component**

## Discussion

The condition was first reported by Atkins et al in 1972. Borgaonkar and colleagues<sup>1</sup> catalogued it as a *genetic syndrome* in 1976. Till date over 60 cases have been reported. All human chromosomes can form a ring chromosome, although they are all very rare, with a combined incidence of only one in 30- 60,000 births.<sup>3</sup> It is Pan-ethnic & non-gender specific condition. It is sporadic except a few with known family history.<sup>4</sup> Most children with Ring 20 syndrome look no different to other children, and are not noticeably short.<sup>5</sup> The

minority who do have an unusual appearance are more likely to have the non-mosaic form of the syndrome.<sup>6</sup> Unusual physical features in this minority can include: Growth delay and being short. A relatively small head, subtle facial features such as down slanting eyes, low placed ears and a small mouth and low muscle tone, so a baby or child feels floppy to hold.<sup>7</sup>

Table 1 shows comparative analysis between the presentation of present case and of Ring 20 chromosome patients.

**Table 1: Features of ring 20 syndrome VS present case**

Feature	Ring 20 syndrome	Present case
Refractory Epilpsy (Generalised Myoclonal Type)	+	+
Mental Retardation	+	+
Speech & Hearing Difficulties	+	+
Short Stature	+	+
Cardiac & Renal Anormalities	+	---
Plagiocephaly	+	+
Microcephaly	+	---
Coarse Facial Features	+	+
Slant in Eyelids	+	+
Hypertelorism	+	+
Cauliflower shaped ears	+	---
Micrognathia	+	---
High arched palate	+	+
Skull views	Normal	Normal
MRI (Brain)	Corpouscallosum, uvula, nodule and cerebellem pyramid hypoplasias.	Focal thinning of corpus callosum at region of posterior one third with splenium.
EEG	Burst of sharply contoured theta activity. Bifrontal spikes and sharp waves	Delta and theta waves in anterior regions at times accompanied by spikes in temporal areas

Definitive diagnosis depends on Chromosome karyotyping. At least 50-100 cells should be cytogenetically analysed to diagnose mosaic ring 20. Management of children with Ring 20 Syndrome needs a multidisciplinary approach. Management of seizures is a clinical challenge. Most cases are Refractory to medical management. Neurosurgery for seizure management is of little or no benefit. Vagus nerve stimulation (VNS) treatment has been reported with a good prognosis in a few cases. The prognosis of condition is assessed based on the degree of seizure control achieved.

## Conclusion

Ring 20 Chromosome Syndrome is a rare genetic disorder with many orofacial and dental abnormalities.

A cytogenetic study should be performed on all patients having epilepsy, dysmorphic features, and/or mentally challenged. The oral physician plays an important role in multidisciplinary approach to the management of these patients.

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