



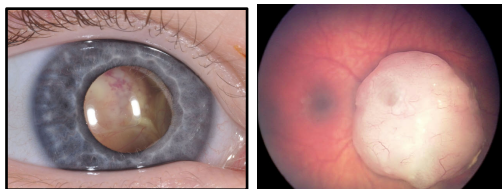
# Airway Challenges in Patients with Retinoblastoma Caused by Chromosome 13q Deletions

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## Introduction

- Retinoblastoma, the most common intraocular childhood malignancy, is caused by the inactivation of RB1 tumor suppressor gene on the long arm (q) of chromosome 13.
- A subset of patients with retinoblastoma have various degrees of 13q deletion involving the RB1 gene.
- Difficult intubation has been noted in this population.



## Method

- Retrospective chart review was performed on retinoblastoma with 13q deletion over 5 years.
- Primary outcome is the best Cormack-Lehane view obtained on direct laryngoscopy. Secondary outcome is the phenotypic features that may contribute to difficult laryngoscopy and intubation.
- Genetic map of the 13q deletion was shown to investigate possible relationship between the degree of genetic deletion, phenotypic features, and difficult laryngoscopy and intubation.

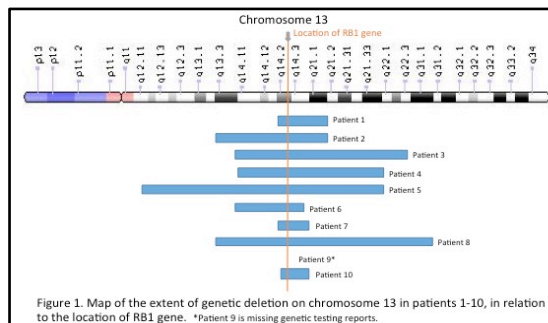


Figure 1. Map of the extent of genetic deletion on chromosome 13 in patients 1-10, in relation to the location of RB1 gene. \*Patient 9 is missing genetic testing reports.

Patient	Eye involved	age at diagnosis	Gene deletion (Mb)	Deletion location	breakpoints or info on deletion	Phenotypic associations	Mask ventilation and LMA	Intubation	Age at 1st intubation
Patient 1	bilateral	22months	7.78	13q14.2 - 13q21.1	47,802,395 - 55,581,949	macrocephaly, global hypotonia, developmental delay	Grade 1 mask. Uncomplicated LMA	Grade 1 view with DL mac2	24months
Patient 2	bilateral	2.5month	17.498	13q13.3 - 13q21.1	35,252,243 - 52,750,209	global hypotonia, severe developmental delay, long narrow face, prominent jaw, small mouth, high palate, asymmetric chest wall	Grade 1 mask	no record	no record
Patient 3	bilateral	DOL 11	NA	13q14.1 - 13q22.3	exons 1 through 27	mild generalized hypotonia, down slanting palpebral fissures	Grade 2 mask. Uncomplicated LMA	Grade 1 view with DL miller 1	4 months
Patient 4	unilateral	17months	28.716	NA	39,456,866-6 8173,640	FFT, "mildly dysmorphic"	Grade 1 mask. Uncomplicated LMA	Grade 1-2a view with Mac 2	18months
Patient 5	bilateral	DOL 7	NA	13q12 - 13q22	no record	short stature, extreme developmental delay, macrocephaly, persistent seizure, b1 hearing loss, malrotation	Grade 1 mask. Uncomplicated LMA	no record	no record
Patient 6	bilateral	14months	NA	13q14	no record	dysmorphic, global delays	Grade 2 mask. Uncomplicated LMA	Grade 1 view with Mac 2	5yr 5 months
Patient 7	unilateral	12months	3.6	13q14.2 - 13q14.3	47,479,099 - 51,042,258	well appearing	Grade 1 mask. Uncomplicated LMA	Grade 1 view with Miller with BURP. History of esophageal intubation.	12months
Patient 8	bilateral	DOL 2	46.4	13q13.3 - 13q31.1	no record	Malrotation, PFO, flat L side head, prominent forehead, eyes slants up, L eye esotropia, hypertelorism, anteverted nares, low set ears and posteriorly rotated, micrognathia, hypotonia	Grade 2 mask. Uncomplicated LMA	Grade 2b view with Miller 1. Grade 1view with Gildescope 1 with BURP. difficult to pass ETT	16months
Patient 9	bilateral	1 month	NA	13q	no record	severe global delay, G tube dependent	Grade 1 mask. Inadequate TV with BURP. Irregularly shaped epiglottis.	Grade 2b with Miller 2	12yrs 6months
Patient 10	unilateral	8months	NA	13q14.3	promoter through exon 27	well appearing, mild global delays, mild macrocephaly and mildly decreased tone	Grade 1 mask. Uncomplicated LMA	no record	no record

## Result

- Ten patients with 13q deletion retinoblastoma were reviewed.
- Two patients had grade 2b view on direct laryngoscopy, and both intubations were noted to be difficult, one of which has the largest confirmed genetic deletion in the cohort.
- Eight patients had craniofacial dysmorphic features.
- Three patients had a grade 2 mask ventilation.

## Discussion

- Incidence of difficult pediatric intubation has been reported: 0.3-9%, however, it is much lower in over 5,000 anesthetics at MSKCC pediatric oncologic ophthalmologic clinic.
- In this study, difficult intubation is noted in 2 out of 10 patients.
- Dysmorphic features are common in this cohort. While dysmorphic features may be a clue, they do not always predict difficult airway.
- Patients with larger degree of 13q deletion may have more difficulty with ventilation, direct laryngoscopy, and intubation.

## Conclusion

Patients with 13q deletion retinoblastoma often need frequent anesthetics for exams and interventions, so anesthesiologists should be cognizant of their potential risk of difficult intubation, which may be related to the degree of genetic deletion and craniofacial dysmorphism.

## Reference

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