

# Bastian Syndrome and the Benefits of Aerodigestive Care

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## Abstract:

Here we describe the unique syndrome of a single family that requires complex pediatric care. There are six cases worldwide of Bastian syndrome. All are within this family, the three siblings, which we will describe, and the mother's two brothers and one sister. Genetically, each child has a practical trisomy of chromosome 13 with an unbalanced translocation of chromosome 6. This leads to a profound clinical presentation requiring complex care. All the siblings have dysfunction in multiple systems. Neurologically, they demonstrate severe developmental delay with intractable epilepsy requiring placement of a vagal nerve stimulator. Each has dysphagia complicated by aspiration and esophagitis requiring placement of a gastrostomy tube and Nissen fundoplication. Their respiratory status is characterized by chronic lung disease with recurrent pneumonia that progresses to non-invasive ventilation and eventual placement of a tracheostomy for two of the three siblings. Each of the siblings has had frequent admissions since birth, many for respiratory and nutritional status. Each admission requires evaluation by specialists including pediatric otolaryngologists, pulmonologists, and gastroenterologists as well as feeding and nutrition expertise. For this rare, poorly characterized syndrome, establishing care in an aerodigestive program has offered coordinated, multidisciplinary care as well as improved communication with the family's medical home and specialists in another state. In doing so, the family was able to have a reduction in hospital visits, a reduction in personal costs, and improved satisfaction with care.

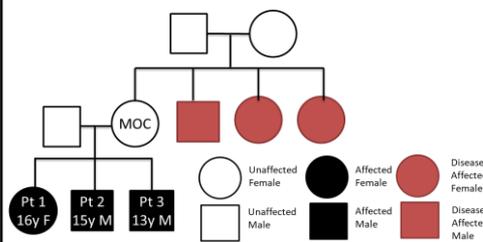
## Objectives:

- 1.) Examine the similar clinical manifestations of this rare genetic mutation
- 2.) Assess benefits in care for family in setting of multidisciplinary aerodigestive clinic

## Methods:

- 1.) Chart review of each of the patients to determine medical comorbidities to define common characteristics of this unique syndrome.
- 2.) Chart review of each of each the patients to assess for changes in number of surgeries and hospital visits pre- and post-establishment in multidisciplinary aerodigestive clinic.

## Bastian Syndrome Family Pedigree



- 3 living siblings with Bastian syndrome of mother of children (MOC), presumed carrier
- 3 siblings of MOC who died at ages of 17y, 19y, and 34y diagnosed with Bastian syndrome
- All diagnosed with practical trisomy of chromosome 13 with an unbalanced translocation of chromosome 6

## Patients



Patient 1) 16 y female



Patient 2) 15 y male



Patient 3) 13 y male

## Anatomy



A.) Airway - chronic aspiration and edema



B.) Esophagus - chronic erythema

## Table 1. Medical Comorbidities

System	Diagnosis	Patient		
		1	2	3
Neurological	Developmental Delay	x	x	x
	Epilepsy	x	x	x
Pulmonary	CLD	x	x	x
	Respiratory failure	x	x	
	Aspiration	x	x	x
GI	Dysphagia	x	x	x
	GERD	x	x	x
	EoE	x	x	x
Cardiac	ASD/ASVD	x	x	
	Clefted mitral valve	x		
Urogenital	Omphalocele			x
	Imperforate Anus			x
Heme	Factor V Leiden	x		

## Table 2. Surgical Procedures

Procedure	Patient		
	1	2	3
VNS	x	x	x
G-tube	x	x	x
Nissen	x	x	x
EGD	x	x	x
Flexible Bronchoscopy	x	x	x
Rigid Bronchoscopy	x	x	x
Tracheostomy	x	x	
AVSD repair	x		x
Mitral valve Replacement	x		
Exploartory Laparotomy		x	
Imperforate Anus Repair			x

## Table 3. Trend in Anesthetic Events

Patient	Pre-Aero	Post-Aero
1	12	4 (2)
2	8	5 (2)
3	9	3 (2)

-Post-Aero time started in aerodigestive clinic. (#) marks combined aerodigestive procedures

## Table 4. Trend in Hospital Admissions

Patient	Pre-Aero	Post-Aero
1	26	4 (2)
2	17	5 (2)
3	6	4 (2)

-Post-Aero time started in aerodigestive clinic. (#) marks combined aerodigestive visits

## Conclusion:

Bastian Syndrome is a rare syndrome unique to this family. It has a broad range of manifestations but almost universally involves intractable epilepsy, chronic dysphagia complicated by aspiration requiring a G-tube and likely eventual tracheostomy. These complex medical diagnosis require multiple specialists and procedures. Early in these patients' care, they had multiple visits to see single providers and for isolated surgical procedures.

Establishing care in a multidisciplinary aerodigestive clinic allowed this family to reduce office visits and hospital admissions. In addition, the program reduced anesthetic events via combined procedures. For patients with complex medical needs like Bastian Syndrome, the combined aerodigestive program offers improved integration and coordination of care to improve patient outcomes and family satisfaction.

## Bibliography

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