Beals Syndrome Affecting a Three-Year-Old: A Case Report

A Hong, K K Patterson

Department of Orthodontics and Pediatric Dentistry, Stony Brook University School of Dental Medicine

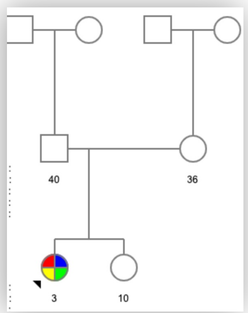
Abstract

Beals syndrome is a rare autosomal dominant connective tissue disorder caused by a mutation in the fibrillin-2 gene (FBN2) on chromosome 5q23.[1-4] FBN2 codes for the fibrillin-2 protein that plays a role in assembling elastic fibers for bone formation and function[4]. Beals syndrome is characterized by multiple flexion contractures, arachnodactyly, severe kyphoscoliosis, abnormal pinnae and muscular hypoplasia[1-4].

The incidence and prevalence of Beals syndrome are unknown as Beals syndrome is phenotypically very similar to Marfan syndrome.[2,3] There is no evidence of shortened lifespan with Beals syndrome.[3]

Clinical Presentation

- Autosomal Dominant Connective Tissue Disorder
  - Mutation in the Fibrillin-2 (FBN2) gene on 5q23[1-4]
- Characterized by
  - Multiple flexion contractures[2-4]
  - Arachnodactyly (long, slender digits)[1-4]
  - Severe kyphoscoliosis[2,4]
  - Abnormal pinnae[2,4]
  - Muscular hypoplasia[2,4]



Mode of Inheritance: Spontaneous Mutation[3]

Flexion Contractures

Kyphoscoliosis

Arachnodactyly

Abnormal Pinnae

Beals syndrome shares traits with Marfan syndrome, those being arachnodactyly (long, slender fingers), Marfanoid habitus (long, thin limbs) and kyphoscoliosis[3] yet it is distinguishable via:

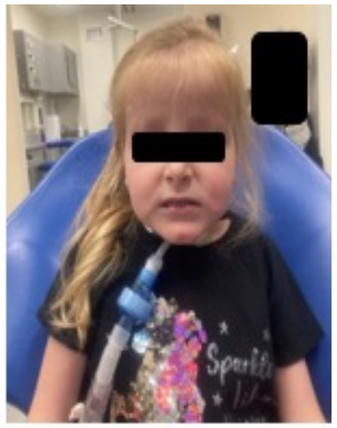
Distinct Traits	Beals Syndrome	Marfan Syndrome
Mutation	Fibrillin-2 gene mutation (5q23)[1-4]	Fibrillin-1 gene (FBN1) mutation (15q15-21.3)[4]
Congenital Joint Contractures	X[1-4]	
Crumpled Ear Appearance	X[1-4]	
Ocular Complications	Ectopia Lentis (Rare)[2,3] Blue sclera, axial myopia, cataract, lens coloboma, ciliary body hypoplasia and glaucoma[2]	Ectopia Lentis (50% of population)[2]
Cardiac Complications	Structural Defects (ventricular, atrial septic defects, patent ductus arteriosus)[2] Aortic Root Dilatation – Stationary[3]	Mitral Valve Prolapse[2,3] Aortic Root Dilatation – Progressive[3]

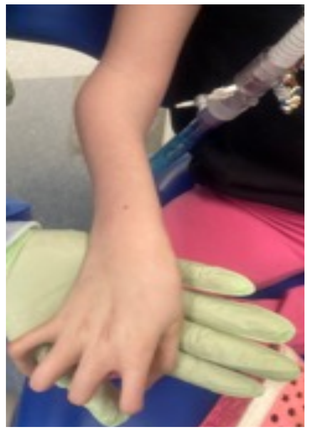
Case Description

A 3-year 8-month-old female of Danish-Norwegian ancestry presented to her dental home at Stony Brook Dental Care Center (Stony Brook, NY) for a periodic dental evaluation. Due to presence of joint contractures at birth, the patient was genetically tested and diagnosed with Beals syndrome confirming a spontaneous mutation in the FBN2 gene with no family history of Marfan syndrome.

The patient presented:
- non-ambulatory
- endotracheal tube
- gastrostomy tube - 50/50 oral/tube feeding
- thoracic rod placement
- aortic root and pulmonary artery dilation
- kyphoscoliosis

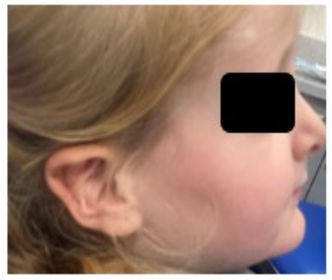
Socially, the patient has limited ability to:
- engage in physical activity
- feed – half of nutrients via gastrostomy tube
- oral hygiene – oral feedings which increase caries risk

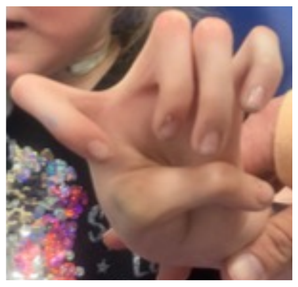




Typical presentation of Beals syndrome:

- Multiple flexion contractures
- Arachnodactyly (long, slender digits)
- Abnormal pinnae





Clinical Relevance

The most significant complication with Beals syndrome is scoliosis and kyphoscoliosis requiring surgical intervention.[2,3] Dental practitioners should recognize these patients are at an increased risk of bacteremia-induced infections and medical consult is obligatory for risk of bacterial endocarditis, need for antibiotic prophylaxis prior to dental treatment and concerns with use of local anesthetics.[5]

Emphasis must be placed on establishing manageable oral hygiene care habits per individual presentation and routine oral health assessments to prevent dental disease and reduce the frequency for antibiotic premedication.[5]

Spinal deformity and kyphoscoliosis may require positioning the patient in a more upright position or in their own wheelchair for patient comfort during dental care.

Contractures and limited mobility must be assessed on an individual basis and manageable oral hygiene may benefit from powered toothbrush and / or interdental brushes with or without modified handles.

Conclusions

Managing a patient with Beals syndrome requires evaluation of the associated spinal deformity, monitoring kyphoscoliosis progression[3] and adjusting the dental environment for greater patient comfort.

Physician consults are necessary regarding spinal issues and associated cardiac defects which may require bacterial endocarditis antibiotic prophylaxis[5].

Ocular issues and joint mobility which affect patient's ability to render self-care including thorough oral hygiene should also be considered on an individual basis.

Appointment duration should be modified for patient comfort, powered toothbrush and/or modified-handled toothbrush or other oral hygiene aids considered and encouragement of dental evaluations with frequency based on individual caries risk assessment.

References

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Disclosures

No funding was required for this case study. Parent and patient permission were granted for photographs.