Prevalence of Tracheostomy Placement in Patients with Skeletal Dysplasia

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BACKGROUND

Skeletal dysplasias (SDs) encompass a heterogenous mix of 461 different disorders. It is known clinically that some SD patients often require early airway interventions. However, there is a paucity of evidence regarding which SD types, how frequently, and at what age these patients require airway procedures. We hypothesized that there is a specific limited subset of SD diagnoses who require frequent and potentially invasive surgical airway management.

METHODS

- Retrospective electronic medical record review of SD patients treated at Seattle Children's Hospital from 1990 to 2022.
- Clinical data warehouse extraction:
  1. 4,306 patients with SD
  2. 313 subjects in this population had an airway procedure
  3. From these patients, 66 had genetically or clinically diagnosed SD
     - Patients in the craniosynostosis syndromes group were excluded
  4. 32 subjects required tracheostomy
- Collected variables included demographics, SD and comorbid diagnoses, airway interventions, and surgical procedures.
- Tracheostomy placement was the primary outcome.

RESULTS

Tracheostomy Prevalence in Subjects with Skeletal Dysplasia by Group.

Group	Group Name	n	Tracheostomy Prevalence
1	FGFR3 Chondrodysplasia	5	0.00%
2	Type 2 collagen	2	50.00%
3	Type 11 collagen	5	40.00%
4	Sulphation disorders	3	66.67%
7	Filamin and related disorders	3	100.00%
8	TRPV4	1	0.00%
9	Ciliopathies with major skeletal involvement	1	100.00%
10	Multiple epiphyseal dysplasia and pseudoachondroplasia	2	0.00%
11	Metaphyseal dysplasias	1	100.00%
14	Severe spondylodyplastic dysplasias	2	50.00%
15	Acromelic dysplasias	1	100.00%
17	Mesomelic and rhizo-mesomelic dysplasias	1	0.00%
18	Bent bone dysplasia	2	100.00%
19	Primordial dwarfism and slender bones	1	100.00%
21	Chondrodysplasia punctata	3	66.67%
24	Other sclerosing bone disorders	2	50.00%
26	Abnormal mineralization	1	0.00%
27	Lysosomal storage diseases with skeletal involvement	5	20.00%
30	Overgrowth (tall stature) syndromes with skeletal involvement	2	0.00%
32	Cleidocranial dysplasia and related disorders	1	100.00%
34	Dysostoses with predominant craniofacial involvement	7	85.71%
35	Dysostoses with predominant vertebral with and without costal involvement	6	33.33%
36	Patellar dysostoses	2	0.00%
38	Brachydactylies (with extraskeletal manifestations)	1	0.00%
39	Limb hypoplasia-reduction defects	4	75.00%
41	Polydactyly-Syndactyly-Triphalangism	2	50.00%

DISCUSSION

- Patients were more often male (56%) with mean age at diagnosis of 2.5 years and mean age at tracheostomy of 1.3 years.
- Overall, 32/66 (49%) patients with SD required a tracheostomy.
- Groups regularly requiring tracheostomy include Filamin and related disorders (100%), Dysostoses with predominant craniofacial involvement (86%), and Limb hypoplasia-reduction defects (75%).
- Feeding difficulties were also common with 81% requiring a feeding tube.
- Craniofacial procedures were performed in 31% of patients.
- A relative limitation of this study was the small sample size in each of the specified SD groups, although it represents the first description of tracheostomy prevalence in many of them.
- Future directions include multi-site recruitment for larger cohorts to continue to evaluate these relationships.

CONCLUSION

Patients with certain SD diagnoses have a higher prevalence of tracheostomy placement. Early identification of these patients may assist in optimizing future airway management strategies.