Case Study:

Collaboration Helps Clinical Team Diagnose Genetic Disorder

SIG 5: Craniofacial and Velopharyngeal Disorders

Summary

An interprofessional practice (IPP) team at a cleft lip and palate clinic developed a treatment plan for a 4-year-old girl with hypernasal speech and dysmorphic features. By working across specialties, the team diagnosed the girl with 22q11.2 deletion syndrome (22q11.2DS) and executed a treatment plan that included speech-language therapy and surgery to create a pharyngeal flap.

Patient Info

ELIZABETH
4-YEAR OLD

Current Diagnosis:
22q11.2 deletion syndrome

Meet The Team

- Audiologist
- Ear, nose, & throat specialist (ENT)
- Geneticist
- Nurse practitioner (NP)
- Plastic surgeon
- Psychologist
- SLP (team facilitator)
- Social worker
- Patient
- Family

Continue for more

Go to Case Rubric

Background

Elizabeth, a 4-year-old girl, had been enrolled in speech-language therapy for 6 months. Concerned about Elizabeth's hypernasal speech, poor intelligibility, and dysmorphic features, the speech-language pathologist (SLP) referred her to an IPP team at the local cleft lip and palate clinic for further evaluation. Elizabeth's parents agreed with the referral.

How They Collaborated

The IPP team members met to discuss Elizabeth's assessment as well as team roles and responsibilities. The team decided that all members would evaluate Elizabeth in a single visit according to their specialty. In addition to evaluations by the SLP, ENT, plastic surgeon, NP, and audiologist, a geneticist would examine Elizabeth, review her family medical history, and determine the need for genetic testing. Afterwards, they would meet as a group to discuss observations and recommendations.

After the evaluations, the team met to share the results and to develop a coordinated treatment plan. The SLP and plastic surgeon found no obvious signs of a submucous cleft palate. The audiologist and ENT found Elizabeth's hearing to be normal. However, during Elizabeth's visit to the clinic, the geneticist explained to Elizabeth's parents the meaning of 22q11.2DS and emphasized the importance of confirming the diagnosis because of other potential implications for her medical care. The lab had drawn Elizabeth's blood for a microarray.

Based on these findings, the team developed and agreed on a treatment plan, which the team facilitator communicated to the family. This included speech-language therapy, a meeting with the geneticist to discuss results of the microarray, and a follow-up procedure with the ENT to confirm a surgical plan.

Outcome

At cleft palate clinic the following month, the results from the microarray confirmed the 22q11.2DS diagnosis. Elizabeth and her parents returned to the cleft lip and palate clinic for a nasopharyngoscopy and to discuss continued treatment. This plan included ongoing speech therapy and surgery to create a pharyngeal flap. Elizabeth and her family also met with the psychologist and social worker to discuss the risk for learning difficulties and to review the mental health concerns associated with her diagnosis.

Ongoing Collaboration

After pharyngeal flap surgery and postoperative assessment, Elizabeth will return at least annually for reevaluation. The team will continue to monitor her outcomes and progress from year to year.

The cleft lip and palate team meets consistently every 2 weeks for a rotating clinic. They maintain communication with each other and with Elizabeth's parents. At the end of their rounds each day, team members debrief about their patients—doing so encourages mutual respect, trust, and collaboration across their disciplines.

Go back to Summary

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Geneticist

Nurse practitioner (NP)

Plastic surgeon

Psychologist

SLP (team facilitator)

Social worker

Patient

Family

Continue for more

History and Concerns

(Share key information gathered from team)

Elizabeth, a 4-year-old girl, was referred to her local cleft lip and palate clinic for concerns with hypernasal speech, poor intelligibility, and dysmorphic features. **Elizabeth** has been enrolled in speech-language therapy for 6 months for articulation. Her current speech-language pathologist (SLP) identified concerns with hypernasal speech and made a referral for further interprofessional (IP) team evaluation. **Elizabeth's** parents agreed with the referral for an IP evaluation.

Assessment Plan

(Determine roles/ responsibilities for evaluation)

The interprofessional cleft lip and palate team members include a SLP (team facilitator), nurse practitioner (NP), audiologist, plastic surgeon, psychologist, geneticist, social worker, and ENT. Team members meet to discuss Elizabeth's assessment needs and together agree upon areas of assessment as well as team roles and responsibilities. The team determines that all members will evaluate Elizabeth in the same clinic, as part of a team visit, and then meet as a group to discuss observations and recommendations. **The assessment plan for the team visit is as follows:**

SLP – Conduct **(a)** perceptual speech evaluation with a focus on evaluation of resonance and any obligatory features of velopharyngeal disorder (VPD); **(b)** standardized articulation assessment; **(c)** oral examination; and **(d)** instrumental assessment, if indicated.

NP – Conduct case history as well as developmental and general medical assessment.

Audiologist – Evaluate hearing; assess middle ear status.

Surgeon – Evaluate anatomy and identify possible etiology of hypernasal speech; collaborate with SLP on velopharyngeal (VP) imaging.

Psychologist – Assess development and behavior.

Geneticist – Evaluate family history and medical history; perform physical exam, and describe any dysmorphic features; determine need for and type of genetic testing.

Assessment Plan

(Determine roles/ responsibilities for evaluation)

Social Worker – Discuss any potential barriers for care; provide community resources.

ENT – Evaluate oral anatomy, middle ear status, airway.

Assessment Results

(Summarize key diagnostic results)

The team meets to share the evaluation results and to develop a coordinated treatment plan:

SLP – The cleft palate team SLP reports that Elizabeth's speech is characterized by glottal stops, glottal co-articulation, nasal substitutions, and final consonant deletions. Parents report typical play and social skills. The SLP judges Elizabeth's speech intelligibility as significantly below age expectations, and parents report that they have a difficult time understanding Elizabeth.

The cleft team SLP indicates Elizabeth has a severe articulation disorder as well as severe hypernasality. The oral exam was unremarkable, with no obvious signs of a submucous cleft palate. Elizabeth failed a language screening, so further formal language assessment is recommended. The cleft team SLP discusses the diagnosis of VPD, the need for VP imaging, and the likelihood of surgical management.

NP – Discusses previous medical history with Elizabeth's parents and the results of a developmental screening. Elizabeth passed in all domains except communication skills.

Audiologist – Reports results of a pure-tone audiogram and tympanometry. Elizabeth displayed responses consistent with normal hearing and middle ear function bilaterally.

Surgeon – Reports that Elizabeth shows no signs of clefting. He discusses the rationale for genetic testing, which is to determine an explanation for Elizabeth's VPD symptoms, dysmorphic features, and the need for surgical management to assess Elizabeth's hypernasality.

Assessment Results

(Summarize key diagnostic results)

Geneticist – Although Elizabeth's family history is unremarkable, the geneticist recommends genetic testing to determine a potential cause for Elizabeth's speech profile, VPD, and dysmorphic features. The lab drew blood for a microarray. He explained to Elizabeth's parents the meaning of 22q11.2 deletion syndrome (22q11.2DS) and emphasized the importance of confirming the diagnosis because of other potential implications for her medical care.

ENT – States that Elizabeth contracted three-to-four ear infections over the past year, although she currently shows normal middle ear function and normal hearing based on audiological testing. He also notes that Elizabeth has no signs suggestive of sleep apnea.

IPP Treatment Plan

(Discuss, reflect, and modify recommendations to develop a coordinated plan)

Based on the reported findings, the IPP team develops and agrees on a treatment plan, which the team facilitator will communicate to the family. **The plan includes the following actions:**

SLP – Continue Elizabeth's current speech-language therapy, targeting articulation. The team recommends a full language evaluation.

Geneticist – Follow up with the geneticist to discuss results of the microarray.

ENT – Have Elizabeth return for a nasopharyngoscopy, so that the ENT can evaluate VP closure for speech and confirm a surgical plan.

Treatment Outcomes
(Discuss results of treatment)

At cleft palate clinic the following month, the results from the microarray confirm the 22q11.2DS diagnosis. **Elizabeth and her parents return to the cleft palate clinic for a nasopharyngoscopy and to discuss continuation of the treatment plan:**

SLP – Speech therapy is ongoing, targeting the sounds /p/, /b/, /t/, /d/, /k/, and /g/. Treatment incorporates nasal occlusion in therapy and in home practice to provide the sensation of oral airflow and pressure. The SLP maintains frequent contact with the team.

Surgeon – The team determines that Elizabeth would benefit from a pharyngeal flap. The surgeon discusses the procedure's risks and benefits. Elizabeth's family agrees to the surgery. The team advises an additional medical workup (i.e., MRI of the neck, kidney ultrasound, cardiac evaluation) because of the 22q11.2DS diagnosis.

Psychologist/Social Worker – Elizabeth and her family meet with the psychologist and social worker again to discuss the risk for learning difficulties and mental health concerns associated with 22q11DS. The psychologist and social worker provide information regarding family support opportunities and community resources. The clinic continues to communicate with Elizabeth's family regarding follow-up needs.

ENT – Conducts the procedure with the SLP and surgeon. The surgeon then discusses the findings and the surgical approach.

After pharyngeal flap surgery and postoperative assessment, Elizabeth will return annually (or more often as needed) for team re-evaluation. The team will continue to engage in reflective practice to monitor her outcomes and progress from year to year.

Team Follow-Up (Determine meetings & communication plan)	The cleft palate team meets consistently every 2 weeks for a rotating clinic and maintains ongoing communication with one another, and with Elizabeth's parents on a regular basis. Team members share decision making through a group debriefing of all patients at the end of the cleft palate clinic rounds for the day, with mutual respect, trust, and collaboration across the various disciplines. Team members resolve conflicts by discussing the pros and cons of various procedures, timing of interventions, and potential risks and benefits until they reach a consensus resolution.

Acknowledgement

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Adriane L. Baylis, PhD, CCC-SLP
Ani Danelz, CCC-SLP
Special Interest Group 5 (SIG 5): Craniofacial and Velopharyngeal Disorders

Citations

American Speech-Language-Hearing Association. (n.d.). Collaboration helps clinical team diagnose genetic disorder. https://www.asha.org/practice/ipe-ipp/case-studies/case-study-2/

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