DOCUMENT RESUME

ED 458 748 EC 308 694

AUTHOR Andrews, Alice E.; Stonestreet, Ruth H.
TITLE Manifestations, Treatment Implications and Speech-Language Consideration in Gorlin Syndrome: A Case Study.
PUB DATE 2001-04-00
NOTE 10p.; Paper presented at the Annual Meeting of the Council for Exceptional Children (80th, Kansas City, MO, April 18-21, 2001).
PUB TYPE Reports - Research (143) -- Speeches/Meeting Papers (150)
EDRS PRICE MF01/PC01 Plus Postage.
DESCRIPTORS Case Studies; College Students; Congenital Impairments; Higher Education; Outcomes of Treatment; *Special Health Problems; *Speech Language Pathology; *Speech Therapy

ABSTRACT

This paper presents a case study of Gorlin Syndrome, also known as Basal Cell Nevus Syndrome, a rare genetic disorder characterized by widespread developmental defects. Criteria for diagnosis are listed, noting the presence of frequent basal cell carcinomas at a relatively young age and multiple cysts of the jaw. Speech and/or language impairments may result from malocclusion, possible cleft palate, and frequent surgeries. The case study is of a 19-year-old female college student who already had 10 surgeries for malignant jaw cysts. Results of hearing, speech, and language tests found normal hearing, expressive and receptive language skills within normal limits, and articulation within normal limits. However, the Dworkin-Culatta Oral Mechanism Examination found numerous abnormalities and weaknesses. Recommendations for speech therapy services to strengthen musculature on one side were made. Decreased muscle flaccidity were noted following the intervention. (DB)

Manifestations, Treatment Implications and Speech-Language Consideration in Gorlin Syndrome: A Case Study

Alice E. Andrews, M.A., CCC-CLP
Ruth H. Stonestreet, Ph.D., CCC-SLP
Department of Special Education and Communication Disorders
Valdosta State University
Valdosta, Georgia

Manifestations, Treatment Implications and Speech and Language Considerations of Gorlin Syndrome: A Case Study

Introduction

Gorlin Syndrome, also known as Basal Cell Nevus Syndrome, is rare with 600 identified cases. It is an autosomal dominant disorder mapped to chromosome 9q22-q31, and allelic loss at this location is common in tumors from Gorlin Syndrome patients (Shimkets, et.al., 1996) This syndrome features widespread developmental defects (Gailani, et.al. 1992).

There are about 100 features of Gorlin Syndrome Major features include basal cell carcinomas, which manifest in cysts of the jaw and skeleton. Odontogenic keratocysts or cysts of the jaw occur in the 1st decade of life, usually about the 7th year and peak in the 2nd and 3rd decades of life (Gorlin, 1987). These cysts are found about 3 times as often in the mandible as in the maxilla. Characteristics facies are present in about 70% of cases and include frontal bulging (Gorlin, 1987). Other features include macrocephaly, rib abnormalities, scoliosis, strabismus and occasional cognitive impairment (Shprintzen, 1997; Gorlin, 1987). Cleft palates occur in about 5% of the cases, and a compensatory articulation disorder may occur if the patient has a cleft palate. Hypernasality may be found secondary to the cleft in a small percentage of patients (Shprintzen, 1997). Hearing acuity and voice are normal,, but language may be delayed or impaired in some cases with central nervous system anomalies and cognitive impairment. Additional features include medulloblastomas, ovarian fibromas, mild hypertelorism, exaggerated length of the mandible and well developed supraorbital

ridges giving the eyes a sunken appearance. Gorlin (1987) reported that there may be fused eyebrows, a broadened nasal root and pouting of the lower lip associated with the exaggerated length of the mandible.

Diagnosis requires 2 major or 1 major and 1 minor criteria. Major criteria includes: (1) more than 2 basal cell carcinomas; (2) less that 30 years of age, cysts of the jaw (odotogenic keratocyst, palmar pits, falx calcification and family history. Minor criteria include: rib or vertebral anomalies, macrocrania, fibromas, medulloblastomas and lymphomesenteric cysts (Gerhardt, 1996).

Of interest to the speech-language pathologist may be the characteristics of a Class III malocclusion (prognathic jaw), high vaulted palate, lateral open bite and possible palatal cleft, possible cognitive impairments, possible language delays or language impairments and the characteristic facial structure. These characteristics may affect speech and/or language production and feeding.

Case Study

A 19-year-old female college student with Gorlin Syndrome was seen at a university speech and hearing clinic. Referral was made by her plastic surgeon from a medical college because of left-sided facial flaccidity. History revealed that the client was diagnosed with Gorlin Syndrome, also known as Basal Cell Nevus Syndrome, at the age of 1 1/2 . The client's father also has this syndrome.

Because of numerous malignant jaw cysts, there have been 10 surgeries as follows:

1981: resection of 2 mandibular cysts

1983: resection of 1 cyst

1985: resection of 1 cyst

1987: resection of 2 maxillary cysts

1988: resection of 1 cyst

1990: resection of 1 cyst

1992: resection of 2 mandibular cysts and ectopic tooth, left maxillary sinus, with iliac bone graft to mandible

1996: resection of 2 cysts, 1 in left maxillary sinus

1998: 2 cysts removed from ovaries

1998: jaw reconstructive surgery

Because of the cysts and surgeries, the client reported that she had abnormal growth of the lower jaw, a Class III occlusal relationship, weak projection of the cheekbones, a large v-shaped deformity at the front of the lower jaw and teeth contact only in the back, giving her an open bite. She reported that she has had severe dental crowding and facial proportions described by her physician as upper midface hypoplasia, malar hypoplasia, an elongated nose and central philtral area with a long midface.

Even though the client was a college freshman, with average to above average grades, a graduate level clinician administered a complete battery of speech and language testing because of reported language and speech problems in clients with Gorlin Syndrome. This clinician was supervised by an Assistant Professor in Speech/Language Pathology who had completed the Deep Pharyngeal Neuromuscular Stimulation training.

Hearing:

A pure tone-hearing test was administered at 25 dB bilaterally. The client passed this test.

Language:

The Test of Adolescent and Adult Language – 3 (TOAL 3) was administered to assess receptive and expressive language abilities.

Listening Vocabulary	50th percentile
Listening Grammar	63rd percentile
Speaking Vocabulary	25th percentile
Speaking Grammar	63rd percentile
Reading Vocabulary	25th percentile
Reading Grammar	63rd percentile
Writing Vocabulary	75th percentile
Writing Grammar	63rd percentile

All of the following scores fell with the average range for her age. Thus it was concluded that her expressive and receptive language skills were within normal limits.

Articulation

The reading portion of the Test of Minimal Articulation Competence (T-MAC) was administered to assess articulation skills. The client obtained a total score of 117 with an articulation index of very mild. The only articulation error noted was a slight distortion of /z/ in the medial position of words. This error was somewhat visually distracting; however, it did not influence her intelligibility. Thus, her articulation was rated within normal limits.

Oral Mechanism Exam

The Dworkin-Culatta Oral Mechanism Examination was administered to examine the oral structures and their function. The following disabilities were noted:

- Left and right jaw and chin area sensation diminished
- Range of lip protrusion reduced – deviation to left
- Imprecise movement of lips – left side appeared weaker
- High narrow arch
- Tongue mildly deviated to left upon protrusion
- Reduced strength of tongue on left side
- Flaccidity of tongue on left side
- Diadochokinetic rate – abnormal syllable stress pattern
- Open bite – left lateral

The client further noted that because of the above-mentioned weaknesses she sometimes had trouble with eating ,especially with biting and then tearing sandwiches. However, she had compensated for these weaknesses by the foods that she chose and slowing down her pace of eating.

Voice and Fluency

Both of these areas were judged to be within normal limits for her age and gender in conversational speech.

Clinical Impressions

The client was found to have muscle flaccidity on the entire left side of her face, including her tongue. Her right side of her face was found to be much stronger than the left.

Prognosis

Stimulability for muscle strengthening of the facial area was judged to be a strength; thus, this intervention was considered appropriate for this client. Because of her self-motivation and cognitive awareness of oral-motor deficits, it was concluded that her prognosis for change was excellent.

Recommendations:

It is recommended that this client receive speech therapy services for one-hour sessions twice weekly. Treatment should focus on:

- Oral-motor excerises specific to the areas that showed flaccidity during the evaluation of the left side of the face
- Icing and massage to strengthen the left side of the face to obtain facial and oral symmetry
- A home program of oral motor exercises twice a day

Intervention:

This client was seen twice a week for one hour at the speech and hearing clinic. A system of icing, massage, and active exercise was initiated. Electrical stimulation (E-stim) therapy was considered as possible treatment. It was contraindicated because of titanium rods placed in the jaw as a result of surgeries.

The client's left facial muscles levator lavii aleque nase, levator labii, zygomatic minor, zygomatic major, obicularis oris, masseter, depressor anguli oris, depressor labii inferioris, risorius/buccinator and mentalis were iced. This procedure involved the application of ice over the target muscle by applying firm, even pressure along the muscle in the direction of the muscle movement. The ice was applied for 3-10 seconds.

This was followed by massage. The clinician applied firm even pressure along the muscle in the direction of the muscle movement.

Finally, oral motor exercises that included the major muscle groups of the face were targeted. The client was directed to imitate the oral movements of the clinician. A large mirror was used to facilitate visual feedback.

After all major muscle groups were iced, massaged and exercised another round of stimulation was performed. Usually each muscle received three episodes of stimulation with a brief rest period between each episode of stimulation.

Results

Muscles of the left side of the face were considered less flaccid. Left side of the mouth was not as turned down. Smile was more symmetrical. At rest, the lips were symmetrical. When at rest, the muscles of cheek and mouth were more taut and judged by the graduate clinician and the client to have more strength. Overall, this intervention was viewed as successful for this client.

Note

This paper describes a single case study on a college student who had been diagnosed with Gorlin Syndrome at the age of 1 ½. Her manifestations of this syndrome may be different from others diagnosed with the same syndrome. The approach described in this paper was specific to this client and may not be appropriate for all clients with Gorlin Syndrome

References

Galani, M.R., Bale, S.J., Leffell, D.J., DiGiovanna, J.J., Peck, G. L., Poliak, S. Drum, M.A., Pastakia, B,. McBride, O.W. and Kase, R. (1992). Developmental defects in Gorlin syndrome related to a putative tumor suppressor gene on chromosome 9.Cell. 111-117.

Gerhardt, M.D.(May 17,1996). Gorlin Syndrome (Basal Cell-Nevus Syndrome). On-line: Virtual Hospital Radiology Resident Case of the Week. http://indy.radiolog,uiowa.edu

, R.J. (1987). Nevooid basal-cell carcinoma syndrome. Medicine. Vol. 66., No. 2. 98-113.

Shimkets, R., Gailani, M.R., Siu, V.M., Yang-Feng, T., Pressman, C.:L, Levanat, S., Goldstein, A., Dean, M., Bale, A.E. (1996). Molecular analysis of chromosome 9q deletions in two Gorlin syndrome patients. American Journal of Human Genetics,59. 417-422.

Shprintzen, R.J. (1997). Genetics, Syndromes and Communication Disorders. San Diego: Singular Publishers.

U.S. Department of Education
Office of Educational Research and Improvement (OERI)
National Library of Education (NLE)
Educational Resources Information Center (ERIC)

REPRODUCTION RELEASE

(Specific Document)

I. DOCUMENT IDENTIFICATION:

Title: Manifestations, Treatment Implications and Speech-Language Considerations in Gorlin Syndrome: A Case Study

Author(s): Alice E. Andrews, M.A., CCC-SLP & Ruth H. Stonestreet, Ph.D., CCC-SLP

Corporate Source:

Publication Date: Presented at CEC April 2001

II. REPRODUCTION RELEASE:

In order to disseminate as widely as possible timely and significant materials of interest to the educational community, documents announced in the monthly abstract journal of the ERIC system, *Resources in Education* (RIE), are usually made available to users in microfiche, reproduced paper copy, and electronic media, and sold through the ERIC Document Reproduction Service (EDRS). Credit is given to the source of each document, and, if reproduction release is granted, one of the following notices is affixed to the document.

If permission is granted to reproduce and disseminate the identified document, please CHECK ONE of the following three options and sign at the bottom of the page.

The sample sticker shown below will be affixed to all Level 1 documents

PERMISSION TO REPRODUCE AND DISSEMINATE THIS MATERIAL HAS BEEN GRANTED BY

Sample

TO THE EDUCATIONAL RESOURCES INFORMATION CENTER (ERIC)

1

Level 1 ↑ ☑

Check here for Level 1 release, permitting reproduction and dissemination in microfiche or other ERIC archival media (e.g., electronic) *and* paper copy.

The sample sticker shown below will be affixed to all Level 2A documents

PERMISSION TO REPRODUCE AND DISSEMINATE THIS MATERIAL IN MICROFICHE, AND IN ELECTRONIC MEDIA FOR ERIC COLLECTION SUBSCRIBERS ONLY, HAS BEEN GRANTED BY

Sample

TO THE EDUCATIONAL RESOURCES INFORMATION CENTER (ERIC)

2A

Level 2A ↑ ☐

Check here for Level 2A release, permitting reproduction and dissemination in microfiche and in electronic media for ERIC archival collection subscribers only

The sample sticker shown below will be affixed to all Level 2B documents

PERMISSION TO REPRODUCE AND DISSEMINATE THIS MATERIAL IN MICROFICHE ONLY HAS BEEN GRANTED BY

Sample

TO THE EDUCATIONAL RESOURCES INFORMATION CENTER (ERIC)

2B

Level 2B ↑ ☐

Check here for Level 2B release, permitting reproduction and dissemination in microfiche only

Documents will be processed as indicated provided reproduction quality permits.
If permission to reproduce is granted, but no box is checked, documents will be processed at Level 1.

I hereby grant to the Educational Resources Information Center (ERIC) nonexclusive permission to reproduce and disseminate this document as indicated above. Reproduction from the ERIC microfiche or electronic media by persons other than ERIC employees and its system contractors requires permission from the copyright holder. Exception is made for non-profit reproduction by libraries and other service agencies to satisfy information needs of educators in response to discrete inquiries.

Sign here, → please

Signature: Alice E. Andrews, M.A., CCC-SLP	Printed Name/Position/Title: Alice E. Andrews, Assistant Professor
Organization/Address: Valdosta State University, 1500 N. Patterson, Valdosta, GA 31698	Telephone: 229-333-5931 FAX: 229-219-1355
	E-Mail Address: aandrews@valdosta.edu Date: 9/20/01

(over)

III. DOCUMENT AVAILABILITY INFORMATION (FROM NON-ERIC SOURCE):

If permission to reproduce is not granted to ERIC, *or*, if you wish ERIC to cite the availability of the document from another source, please provide the following information regarding the availability of the document. (ERIC will not announce a document unless it is publicly available, and a dependable source can be specified. Contributors should also be aware that ERIC selection criteria are significantly more stringent for documents that cannot be made available through EDRS.)

Publisher/Distributor:
Address:
Price:

IV. REFERRAL OF ERIC TO COPYRIGHT/REPRODUCTION RIGHTS HOLDER:

If the right to grant this reproduction release is held by someone other than the addressee, please provide the appropriate name and address:

Name:
Address:

V. WHERE TO SEND THIS FORM:

Acquisitions Coordinator
ERIC Clearinghouse on Disabilities and Gifted Education
1110 North Glebe Road
Suite 300
Arlington VA 22201-5704

However, if solicited by the ERIC Facility, or if making an unsolicited contribution to ERIC, return this form (and the document being contributed) to:

ERIC Processing and Reference Facility
1100 West Street, 2nd Floor
Laurel, Maryland 20707-3598

Telephone: 301-497-4080
Toll Free: 800-799-3742
FAX: 301-953-0263
e-mail: ericfac@inet.ed.gov
WWW: http://ericfac.piccard.csc.com

EFF-088 (Rev. 9/97)