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Case report

Evaluation for language and speech development in Kabuki make-up syndrome: A case report

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ABSTRACT

Kabuki make-up syndrome (KMS) is defined as a rare syndrome with mental retardation, growth deficiency and multiple anomalies of unknown cause. Cases have a characteristic facial appearance of broad and low auricles, wide forehead, broad and flattened nose root. In this article, 4-year, 10-month-old boy with speech delay reported due to characteristics of the facial appearance is considered as KMS, a rare syndrome. Otological, audiological and developmental evaluation of the patient consisted of six parameters. (1) ENT examination: normal. (2) Audiological findings were at normal hearing levels. (3) General development (according to the Denver II test), was normal. (4) Language and speech development: receptive language development was 2 years more than the chronological age. However in expressive language development, there was 5-month delay which was likely to complete by training in a short time. (5) The development of speech sounds and articulation: the delay is observed in CVC words. Speech sounds of /l/, /t/, /r/ and /g/ were evaluated as "distorted expression". He used /v/ instead of /l/; /y/ instead of /r/; /d/ instead of /g/. (6) Observations: diffident, deficit in self-confidence, and difficulty in communicating have been observed.Unlike the cases in literature, mental retardation, growth deficiency and learning difficulties were not found in our case with KMS. Normal findings were obtained in five parameters. Only the /l/, /r/ and /g/ speech sounds were expressed as distorted. For our case, we planned to follow him in the future to see whether developmental and central auditory processing disorders will occur or not.

1. Introduction

Kabuki syndrome (KS) or Kabuki make-up syndrome (KMS) (Niikawa–Kuroki syndrome) is a multiple malformation/mental retardation syndrome that was described initially in Japan but is now known to occur in many other ethnic groups. It is characterized by distinctive facial features (eversion of the lower lateral eyelid, arched eyebrows with the lateral one-third dispersed or sparse, depressed nasal tip, and prominent ears), skeletal anomalies, dermatoglyphic abnormalities, short stature, and mental retardation. A number of other manifestations involving other organ systems can aid in the diagnosis and management of KMS [1].

Kabuki make-up syndrome is most commonly seen in Japan; and reported from different geographic regions in the world. To date, only 350 cases have been identified in the literature; and it is expressed as 1/32.000 incidence in Japan [2,3]. Tsukahara et al. [4] reported dominant inheritance in KMS. They also reported empty sella, maxillary hipoplasia and a lot of pits in the skin. Burke and Jones [5] presented eight cases of Kabuki syndrome in non-Japanese patients. Clinical features include a characteristic facies, developmental delay, musculoskeletal abnormalities, and dermatoglyphic differences. The phenotype appears to evolve over time making the diagnosis difficult in infancy. The progressive changes in the facial characteristics as well as the musculoskeletal problems suggest an underlying defect of the connective tissue. They reported that the syndrome appeared to be more common in the non-Japanese population than previously appreciated, particularly in the cleft palate population.

Yavaşcan, et al. [6] presented 5-year-old girl, speech disorder, hyperemia in the eye, recurrent discharge in the ear, cleft palate, atrial septal defect (ASD) and membranous ventricular septal defect (VSD) were seen. Wide forehead, eyebrows arched upwards,

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bilateral ectropion in the eyes, nose root's width, lower located ears and nape hairline and a high palate were detected in the examination.

Upton et al. [7] investigated speech patterns associated with Kabuki syndrome. All the six children had a history of delayed speech and language acquisition and were receiving speech services. All individuals had articulation errors and abnormal oral resonance, which appeared to be due to poor oral-motor coordination and hypotonia and were not felt to be due to structural abnormalities such as velopharyngeal insufficiency, dental malocclusion, or cleft palate. An intriguing finding, noted in the two individuals followed from childhood into adolescence with serial speech evaluations, was that pitch, loudness, and prosody did not mature over time and what was age appropriate performance at younger ages became inappropriate in adolescence. They concluded that distinctive speech characteristics with a lack of normal maturation during childhood can be added to the extensive list of clinical features associated with the Kabuki syndrome and hopefully will lead to improved speech/language treatment for individuals with this syndrome.

Defloor et al. [8] reported speech language development of Kabuki syndrome. According to their findings, expressive language abilities were impaired. Poor morphosyntactic abilities were consistently demonstrated. Lexical and pragmatic difficulties were also present, whereas phonological development was less often affected.

In this article, we reported otological, audiological and developmental features of the patient with KMS which was referred to our clinic for language and speech evaluation for the complaint of language delay.

2. Case report

The 4-year and 10-month-old male patient was seen for the evaluation of language and speech development in Hacettepe University, Faculty of Medicine, Division of Audiology and Speech Pathology. No relationship was present between the parents.

The physical and ENT examinations revealed typical facial appearance of the patient for KMS: auricles were low and prominent. There were also light trigono-cephaly, blue sclera, abnormal dentition, high-arched palate, prominent nasal root, werted palpebral lateral part (Fig. 1); short neck, low hairline (Fig. 2). Fetal pads were present in fingers (Fig. 3) and breast heads were splitted. He had also ASD which was improved at the current time and spina bifida at birth (knowledge was present in hospital file of the patient). Father of the child has also similar facial appearance, without any mental and developmental delay.

In developmental history of the child:

- keep head upright at 1–1.5 months,
- assisted sitting at 7–8 months,
- non-assisted sitting at 10 months,
- walking at 16–18 months,
- saying a few words at 18 months, and
- first sentence was said at 4 years.

Otological, audiological and developmental evaluation of the patient consisted of six parameters:

(1) ENT examination: normal.
(2) Audiological findings: the patient was evaluated by pure tone audiometry, tympanometric tests, acoustic reflex, transient evoked otoacoustic emission (TEOAE), distortion product otoacoustic emission (DPOAE), auditory brainstem response (ABR). Audiological findings were found as normal hearing levels.

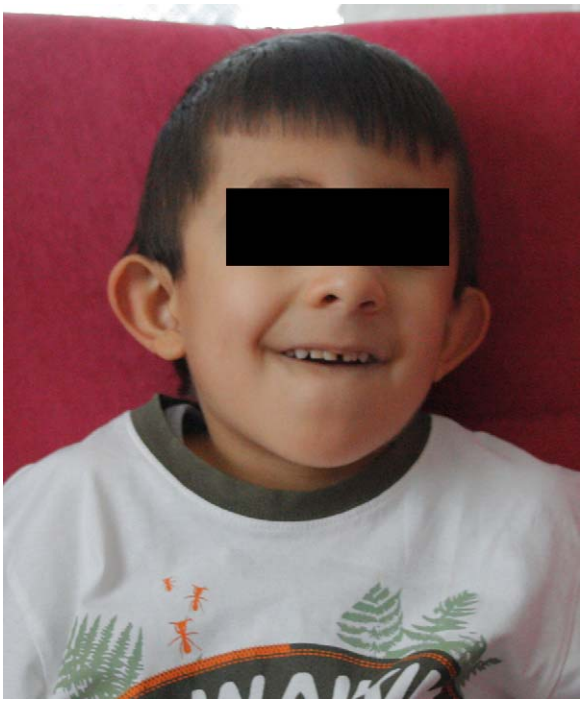

Fig. 1. Antero-posterior view of the child. Auricles were low and prominent. There were also light trigono-cephaly, blue sclera, and abnormal dentition.

(3) General development is evaluated by:
   (a) Ankara Developmental Scanning Inventory [9] had been applied (knowledge was present in hospital file of the patient).
   - General development: 4 years 6 months.

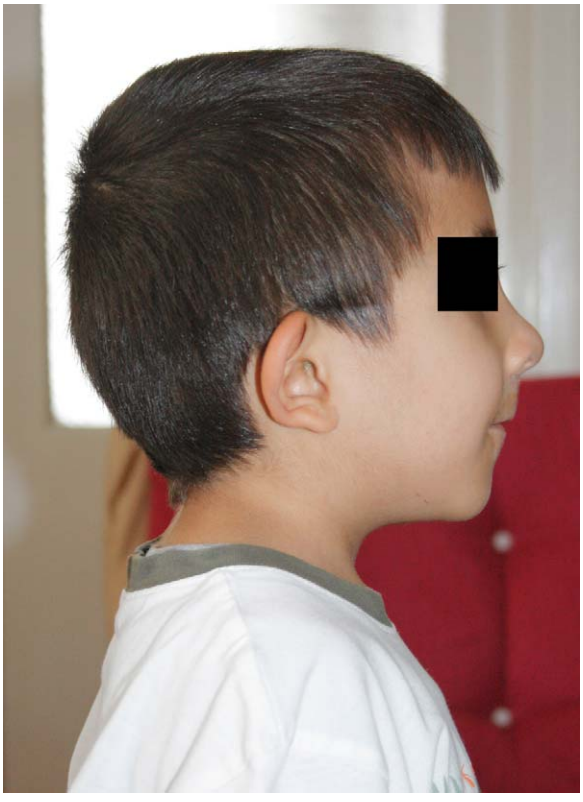

Fig. 2. Right-lateral view of the child, showing short neck, low hairline and blue sclera.

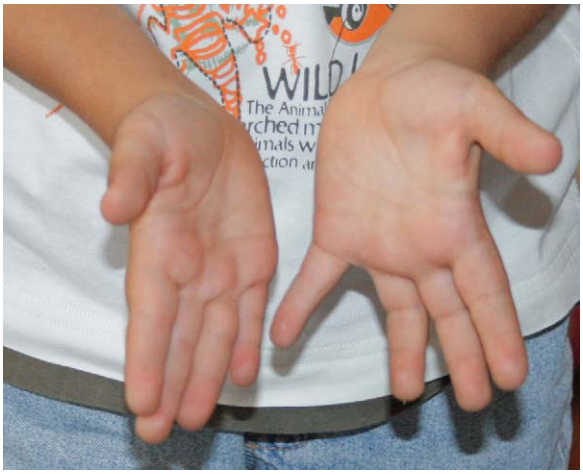

Fig. 3. Fetal pads were present in fingers.

- Language-cognitive development: 4 years 6 months.
- Fine motor: 5 years (above his age).
- Rough motor: 3 years 6 months.
- Social skills and self-care skills: 4 years 10 months.

These test results were compatible with the overall development. The child was impressed as compatible with his coevals.

(b) Denver II test (Anlar and Yalaz [10]): overall development was normal (4 years 11 months). As a result of Denver II test, delay in language development was found. Fine and rough motor development; and personal–social skills were found as normal.

(4) Language and speech development: receptive and expressive language development was assessed by Language Scale 4 test [11]. Chronological age was 4 years 10 months at the date of test.
- Receptive language: 5 years to 5 years 5 months (60–65 months).
- Expressive language: 4 years to 4 years 5 months (48–53 months).

According to the Language Scale 4 test, receptive language development was 1-year more than the chronological age. However in expressive language development, there was 5-month delay which was likely to complete by training in a short time.

(5) The development of speech sounds and articulation: It was assessed by Speech Sounds Development Test (SSDT) [12]. The delay is observed in CVC words. Speech sounds of /l/, /t/, /r/ and /g/ were evaluated as "distorted expression". He used /v/ instead of /l/; /y/ instead of /r/; /d/ instead of /g/.

(6) Observations: diffident, deficit in self-confidence, and difficulty in communicating have been observed.

Child's father gave written and signed permission for using all data and photographs of the child for scientific aims (publications and presentations).

3. Discussion

In literature, mental retardation and developmental delay are identified in children with KMS [3,13–16]. However, in our study, developmental delay and mental retardation were not present, unlike the literature. Our results show that mental retardation and developmental delay may not be in certain diagnostic criteria of a KMS.

Our case with KMS, child's social contact with parents and familiar people was normal. At first, he was diffident; but when orientated, his contact was much better. He used pragmatic language (greeting, good day, thank you, etc.). Our findings show that KMS does not affect the social communication in fundamental level. In other words, the child uses pragmatic skills in social communication at basic level. But these basic-level used pragmatic skills do not reflect that the child does not have any problems in pragmatic areas. He still has troubles in usage of pragmatic language use in daily life. His family is too protective; therefore his communication with peers and environment was not supported sufficiently by his family. As a result, there are great problems in usage of pragmatic skills in social communications.

In our case, language and speech development was evaluated. Especially receptive language development was 1 year more than the chronological age. However in expressive language development, there was 5-month delay. Examples for language delay are: "Responds to where question; to complete analogues; names of objects when the object is described". Expressive language delay may be related to communication disorders and environmental deprivation. Because, the child's family is over protective.

He has some speech sound problems. The delay is observed in CVC words. In speech sounds, in some sounds, there were disorders (/l/, /r/, /g/); and in some sounds, there were substitutions. He often makes speech errors, and has phonological and articulation disorder. Some sounds were substituted, such as /v/ was used instead of /l/.

In our case, language and speech sound development results were similar to Van Lierde et al.'s [17] case, in which the case of a girl aged 3 years 8 months with Kabuki make-up syndrome is reported. At presentation, she had normal cognitive functioning, and she also had a history of otitis media, a submucous cleft palate, and some hypotonia. Language testing showed normal receptive skills and good expressive vocabulary but poor morphosyntactic abilities. Speech analysis showed that she was capable of producing most of the sounds of her native language but demonstrated high variability in the production of the sounds. In addition, she inconsistently simplified words by the application of several phonologic processes. Possible explanations for the communication problems demonstrated are discussed.

For our case, the child's vocabulary is not rich. But we have not seen it as pathological. Because, his family is over protective. The words, used in family and child communication, are certain, such as, "eat more", "drink milk", "go to bed", etc. which contain limited words. In our case, the child cannot have opportunity to play with peers and experience the life interactively. Because of all these reasons, we do not assess the child's not-rich vocabulary as pathological. Although the communication with environment is provided; and he had received warnings and stimuli enough; and he had no rich vocabulary; only in these conditions, we can say that vocabulary development is weak. For our case, it can be said that the child have normal morphosyntactic skills, but specific morphosyntactic difficulties emerged in production.

Our findings showed that this syndrome did not affect the language, but affected the development of speech sounds. This impression was as a result of observations in child's educational follow-up. Speech sound development may be increased with speech therapy and his families' applications according to our directions. Also, some normal (without syndrome) young children often make speech errors, and have phonological and articulation disorders in early ages. But these disorders may be improved in the following years.

Conflict of interest

The authors declare that there is no conflict of interest.

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