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A Case Report: Hallermann-Streiff Syndrome

CASE REPORT

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ABSTRACT

Hallermann-Streiff is a rare syndrome characterized by multiple congenital anomalies, especially in the head and face. It was first described by Hallermann in 1948 and later by Streiff in 1950. Patients have a bird-like face, skin atrophy, dwarfism, various eye abnormalities, and hypotrichosis. In the case admitted to our hospital with complaints of fever, cough, and failure to thrive, we detected microphthalmia, bilateral leukocoria, cataract, eyebrow hypotrichosis, a prominent nose, and depressed nasal root. The patient was diagnosed with Hallermann-Streiff syndrome. This case was presented because of its rare occurrence in the literature.

Key words: Hallermann-Streiff Syndrome, cataract, microphthalmos

INTRODUCTION

Hallermann-Streiff syndrome (HSS) is a syndrome including mandibulo-facial anomalies described by Hallermann in 1948 and by Streiff in 1950 (1). Although familial cases have been reported in low numbers, HSS often presents with sporadic mutations (2, 3). The characteristic clinical features of the syndrome are a bird-like face, mandibular and maxillary hypoplasia, failure to thrive, bilateral microphthalmia, congenital cataract, dental anomalies, skin atrophy, and hypotrichosis (4, 5). Patients usually die due to recurrent respiratory tract infections, sleep apnea syndrome, and respiratory arrest (2). In this study, a 7-month-old case diagnosed with rarely seen Hallermann-Streiff syndrome is presented.

CASE REPORT

A 7-month-old baby boy was admitted to our clinic with complaints of fever, cough, and failure to thrive. The patient was the son of a 40-year-old healthy father and 36-year-old healthy mother as the 6th living child from the 9th pregnancy. Second-degree consanguinity was reported between the parents. It was also reported that the mother had not been followed during pregnancy. Moreover, the baby had not been taken to a hospital for medical controls, and also, he had not been vaccinated. In his physical examination, his weight was 4750 gr (<3 percentile), height was 59 cm (<3percentile), head circumference was 40 cm (<3percentile), body mass index was 13 (<3percentile), and upper/lower segment ratio was 1.45. Microphthalmia, bilateral leukocoria, eyebrow hypotrichosis, broad anterior fontanel (6x2 cm), a prominent nose, depressed nasal root, high-arched palate, low-set ears, and small mouth were detected in the patient (Figure 1).

In the respiratory system examination of the patient, rhonchi and rales were found. The abdomen was free of organomegaly. The results of the cardiovascular system and genitourinary system examination were normal. The values of the complete blood count; serum electrolytes; liver and kidney function tests; growth hormone, thyroid, and parathyroid hormone levels; and immunoglobulins (IgA, IgG, IgM, and IgE) were found to be normal (Table 1). In the radiological examination, bronchopneumonia infiltration was detected on the right side in the chest radiography. Minimal dilatation was observed in the lateral ventricles and third ventricle in the brain tomography. No pathology was found in the whole-body radiographies. His echocardiography examination result was normal. In a chromosome analysis carried out with the GTG-banding technique on a peripheral blood culture, no number or structural anomaly was observed. The case was diagnosed with Hallermann-Streiff syndrome, considering the clinical findings. The patient was followed up by the ophthalmic clinic due to the eye-related symptoms.

DISCUSSION

Hallermann-Streiff is a rarely seen syndrome characterized by clinical features, and its etiology remains unknown (1, 2, 6). In the literature, cases with a familial medical history, a case whose mother had a rubella infection in pregnancy, a case whose mother used various medications in the first trimester, and sporadic cases have been reported (6-9). A 4p anomaly was detected in a patient, and 4q monosomy and partial 14q trisomy were found in another

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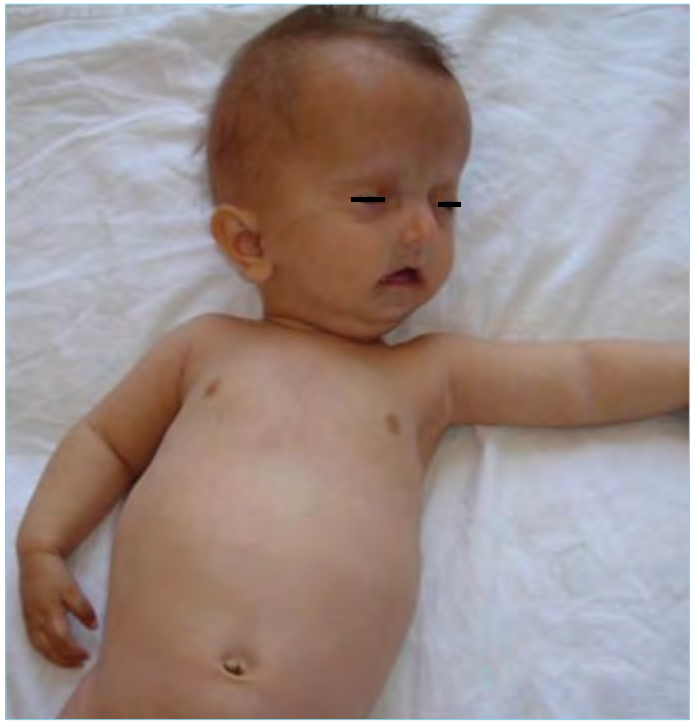

Figure 1. Typical facial appearance (microphthalmia, eyebrow hypotrichosis, prominent nose, depressed nasal root, low-set ears, and small mouth) and anatomy of the patient
(The parents of the patient were informed before photographs were taken, and written consent was obtained.)

patient (3). In spite of these findings, the gene causing HSS has not been identified yet. A mutation in gap junction alpha-1 protein (GJA1) was found for oculodentodigital dysplasia (ODDD), which occurs with more severe clinical findings compared to HSS (3, 10). A similar mutation was reported by Damiano et al. in a patient diagnosed with HSS, but this mutation was not found in any other case with HSS (3, 11). Ultimately, a mutation in the GJA1 gene is controversial for patients with HSS. A skin biopsy revealed changes in collagen and elastin and a decrease in mucopolysaccharide (1). In our case, we found that the mother had not used any medication during her pregnancy and had no infection. However, she had not seen the doctor for follow-up controls during pregnancy. In the chromosomal analysis of the patient, no number or structure anomaly was observed. There was no other individual diagnosed with HSS in his family. Therefore, his disease was thought to have occurred due to a sporadic mutation.

The patient had 5 of 7 diagnostic criteria developed for Hallermann-Streiff syndrome (bird-like facial appearance and prominent nose; abnormal odontogenesis; hypotrichosis; skin atrophy, especially on the nose; congenital cataract; bilateral microphthalmia, and proportional dwarfism) (1). It is stated that the diagnosis of HSS should be reviewed in the absence of eye-related findings (cataract and microphthalmia) (6, 7). In the literature, it is emphasized that eye-related findings should be prioritized in the treatment, because they may lead to blindness (4). Our case had both cataract and bilateral microphthalmia. He was followed up by an ophthalmic clinic. The patient had a broad forehead, high-arched palate, low-set ears, thin and brittle hair, small mouth, and broad anterior fontanel, in addition to the typical bird-like facial appearance. These findings were consistent with the findings in the literature (1, 6). In the literature, it is mentioned that natal tooth, cleft palate, syndactyly, and congenital cardiac diseases (atrial septal defect, ventricular septal defect, patent foramen ovale, etc.) can be seen in some HSS cases (4, 6, 12). Cleft palate, syndactyly, and congenital cardiac defects were not found in our case. A history of natal tooth at birth was not found.

Table 1. Laboratory results of the patient

	Laboratory results	Normal reference values
White blood cell count (/mm^3)	8.6	4-11
Hemoglobin (g/dL)	11.8	11.5-15.5
Hematocrit (%)	36	35-45
MCV	80	70-86
Thrombocyte count (/mm^3)	312	150-400
Glucose (mg/dL)	83	60-100
Urea (mg/dL)	23	0-50
Creatine (mg/dL)	0.3	0.3-0.7
Sodium (mEq/L)	139	135-145
Potassium (mEq/L)	4.2	3.5-6
Chlorine (mEq/L)	98	98-106
Calcium (mg/dL)	9.5	8.8-10.8
Phosphorus (mg/dL)	5	3.8-6.5
AST (U/L)	35	5-45
ALT (U/L)	36	5-45
Free T4 (ng/dL)	1	0.61-1.12
TSH (mIU/L)	4	0.7-6
Parathormone (pg/mL)	10	<10-65
GH (ng/mL)	1.04	0-6
IgA (mg/dL)	75	11-106
IgM (mg/dL)	44	33-126
IgG (mg/dL)	198	172-1069
IgE (mg/dL)	169	0-230

ALP: Alkaline phosphatase, ALT: alanine aminotransferase, AST: aspartate aminotransferase, GH: growth hormone, MCV: mean corpuscular volume, TSH: thyroid-stimulating hormone

Symmetrical growth failure existed in the patient. However, hypoparathyroidism and osteopenia, which are reported in the literature, were not observed in this case with low growth hormone (11, 13). Although a low level of insulin-like growth factor was reported in similar cases, the level of insulin-like growth factor was not investigated in our case (11). It is stated in the literature that pulmonary infection, apnea, and respiratory arrest can frequently develop due to the anatomical abnormality of the respiratory tract (mandibular hypoplasia, microstomy, etc.), and there may be some cases requiring tracheostomy because of respiratory distress (2, 5, 6, 8, 9, 12).

In our case, the patient had a pulmonary infection lasting for 1 month at the time of admission to the clinic. In the literature, a 3-month-old case with a difficult intraoperative intubation was reported (12). Therefore, intubation should be performed carefully, considering the possibility of respiratory tract anomalies.

CONCLUSION

The complex problems of cases with rarely seen Hallermann-Streiff syndrome necessitate the follow-up of these patients in an experienced health center.

Informed Consent: Written informed consent was obtained from patients' parents who participated in this study.

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