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Case Report
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TRISOMY OF 1q31.3q42.12 CHROMOSOME: CASE REPORT

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Abstract: Chromosome 1q duplication is one of the rare congenital anomalies accompanied by numerous visceral organ anomalies, dysmorphism, and psychomotor retardation. Our case is an 8-year-old male patient with 1q31.3q42.12 trisomy. The patient was brought to our rehabilitation center because he couldn't able to go up and down stairs and slopes without support also he couldn't run and jump. The patient receives physical therapy service for kyphotic posture, speech therapy for speech and drooling problems, and special education support for mild cognitive impairment. Our aim is to describe the dysmorphic features and posture examination of an 8-year-old male patient with chromosome 1q31.3q42.12 trisomy. In lateral, anterior and posterior posture examination, various anatomic and dysmorphic problems were observed on the patient. To improve the life quality and comfort of the patient, it should be prepared necessary treatment plans for this patients.

Keywords: Dysmorphism, Genetic, Trisomy

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1. Introduction

Chromosome 1q duplication is one of the rare congenital anomalies accompanied by numerous visceral organ anomalies, dysmorphism, and psychomotor retardation. While abnormalities in the long arm of chromosome 1 are associated with both hereditary disorders and neoplastic diseases, partial trisomy 1q abnormality is rarely seen. The distal half of the long arm of chromosome 1 is often involved (Pettenati et al., 2001; Chan et al., 2002; Güneş et al., 2005).

Findings such as microretrognathy, high palate, short neck, flexion contracture in extremities, finger deformities (mallet finger), and kyphoscoliosis are the most common dysmorphic features in 1q duplications (Pettenati et al., 2001; Nowaczyk et al., 2003; Bükülmez et al., 2009). Also, in 1q duplication, intrauterine and postnatal growth retardation, solid tumors, invasive breast carcinomas, cervical cancers, renal cell carcinoma, sarcomas, and hepatocellular carcinomas are frequently recorded (Bueger et al., 2000; Matthews et al., 2000).

Duplication 1 (q24q ter) and 1 (q25q ter) are the type of duplication with the most severe anomalies, while duplication 1q (32-q sweat) is with the shortest life span (Van Haelst et al., 2002).

This study aims to describe the dysmorphic features and posture examination of an 8-year-old male patient with chromosome 1q31.3q42.12 trisomy.

2. Case Report

Our case is an 8-year-old male patient born in 2012 with 1q31.3q42.12 trisomy. Prader Willi Syndrome was pre-diagnosed 1 month after birth of the patient. After 5 months, Prader Willi Syndrome was ruled out with the methylation analysis performed on the patient. The diagnosis of the patient was made as 1q31.3q42.12 trisomy as a result of chromosome analysis performed in the Hacettepe University Genetics Unit in 2014.

The patient was brought to our rehabilitation center because he couldn't able to go up and down stairs and slopes without support, run, and jump; also to receive physical therapy service for kyphotic posture, speech therapy for speech and drooling problems, and special education support for mild cognitive impairment.

In the anamnesis taken from the family, the mother and father did not have a consanguineous marriage. The mother had a miscarriage in her first pregnancy due to an ectopic pregnancy. She gave birth to a healthy male child by normal birth in the second pregnancy, who was 36 weeks old and is now 16 years old; and abortions have occurred in the next 2 pregnancies. When the mother was 36 years old, she was followed up with suspicion of fetal abnormality in her fifth pregnancy, and our patient was born by cesarean at 36 weeks and 5 days due to fetal distress with a weight of 2700 g. The mother has prenatal polyhydramnios in both her born children. None of her relatives has experienced such a case before.

As soon as our patient was born, it was noticed that his color was cyanotic and he was taken into the intensive

care unit. In further examinations, laryngomalacia was detected. When the formula feeding was started, it was observed that the baby had difficulty swallowing. Therefore, the baby was started to be fed with a nasogastric (NG) tube and remained in the incubator for 40 days. NG was kept until 6 months of age, after 6 months he was fed with liquid foods until 2 years of age, after 2 years of age, he was fed with mashed foods, and with 3 years of age, it was switched to solid food.

Postnatal jaundice was detected in the patient and he received phototherapy for a while. Due to congenital hypothyroidism, Euthyrox was used for 6 months and now his values are within the normal range, and he is not using any medication. Our patient underwent unilateral undescended testicular surgery when he was 6 months old, and his brother also had undescended testicle findings. At the age of 18 months, our patient underwent Patent Ductus Arteriosus and Atrial Septal Defect surgeries due to heart problems. With the examination performed in the pediatric urodynamics outpatient clinic in 2019, it was observed that the right kidney was in normal anatomic location but slightly small. Their contribution to total kidney function was calculated as 58% for left and 42% for right. The result of the hearing test was normal. Our patient wears glasses due to hypermetropia at 4.5 degrees in the right eye and 8.5 degrees in the left eye, and astigmatism. Loss of transparency was observed in the cornea examination. The patient is currently not using any medication.

In the normal motor development history of the patient, it was noted that he held his head at the age of 6 months, switched to supported sitting at the age of 9 months, and began to sit unsupported around 1 year old. He started walking around the age of 18 months and started walking independently after the age of 2.

In the lateral posture examination we performed on the patient, short neck, shoulder head protraction, increased thoracic kyphotic posture, flexor posture in the hip, knee, and elbow joint; posterior pelvic tilt and pes planus was observed (Figure 1).

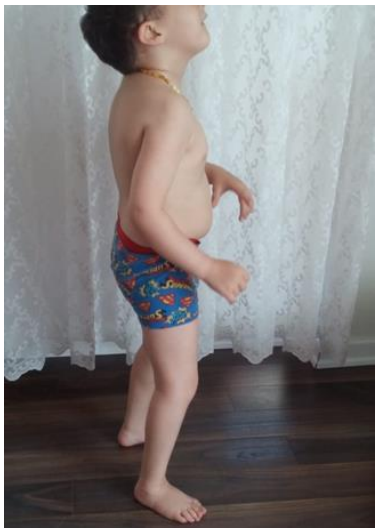

Figure 1. Lateral postural stance.

In the anterior and posterior posture examination, there is the onset of C scoliosis on the right side of the upper thoracic region, abduction and external rotation in the lower extremities, hallux valgus in the big toes, and the trigger finger onset on the thumb of the hand (Figure 2 and Figure 3).

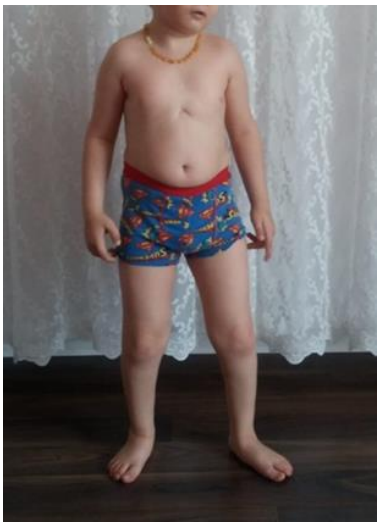

Figure 2. Anterior postural stance.

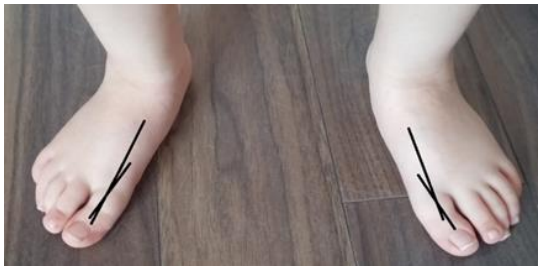

Figure 3. Hallux valgus deformity of the foot.

Other dysmorphic findings in the patient are bilateral microphthalmia, microretrognathia, reduction in mouth opening, narrow palate, and small ears. Also, hypoplasia in the external genital organs was noted (Figure 4).

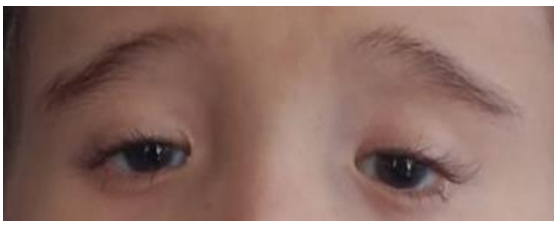

Figure 4. Bilateral microphthalmia.

In our rehabilitation center, we makes stretching for the patient's muscle shortness. Muscle strengthening is used for low muscle tone. Scoliosis, balance and coordination exercises are practiced.

3. Discussion

Chromosomal abnormalities include situations such as numerical and structural anomalies; or marker

chromosomes, where both are together. Chromosomal abnormalities are seen in approximately 1/200 of the newborn. This rate increases even more in preterm births and miscarriages (Yirmibeş Karaoğuz, 2007; Çınar Kuşkucu, 2010). There are also abortions in the pregnancy history of the mother, and our patient was born when he was 36 weeks and 5 days old.

Currently, chromosomes can be obtained by culturing samples such as peripheral blood, bone marrow, amniotic fluid cells, buccal mucosa, skin fibroblasts. Structural and numerical anomalies of chromosomes are determined, which are obtained from these tissues. Generally, chromosome analysis is performed by culturing peripheral blood tissue (Nowaczyk et al., 2003; Zamani, 2007; Zamani, 2013). Our patient was diagnosed as Prader Willi after birth, however, was ruled out after 5 months. Thereafter, a diagnosis of 1q31.3q42.12 trisomy was made in the chromosome analysis performed by culturing peripheral blood tissue.

Microretrognathy, high palate, short neck, flexion contracture in extremities, finger deformities (mallet finger), kyphoscoliosis are the most common dysmorphic features seen in 1q duplications (Pettenati et al., 2001; Nowaczyk et al., 2003; Bükülmez et al., 2009). The results of examinations performed on our patient are consistent with these dysmorphic features.

It can be considered that the tumor suppressor gene is localized on the 1q chromosome. Studies have shown that allelic losses on 1q are frequently associated with many cancers such as breast cancer, medulloblastoma, thyroid cancer, sporadic insulinoma, colorectal carcinomas, and esophageal cancers (Yang et al., 2005; Fromont et al., 2007; Zhou et al., 2008).

In cases with 1q duplication, some cases with ambiguous genitalia and genital hypoplasia have been reported as well (Van Haelst et al., 2002). Genital hypoplasia was also encountered in our patient and testicles are palpable.

In accordance with dysmorphic definitions and the postural examinations we performed on our patient, we found that the patient had motor development retardation, anatomical, biomechanical and multi-organ problems. If the increased kyphotic posture and scoliosis observed as a result of postural examination are not controlled with the necessary treatments, it could cause serious respiratory, aesthetic, and musculoskeletal problems in the future.

The pes planus and hallux valgus in the foot structure, and the flexor posture in the lower extremity can cause pain and deformations in the anatomical structure of the patient by causing biomechanical alignment problems. The upper extremity flexor posture and trigger finger onset in the patient can reduce the functionality and cause limitations in living conditions.

Surgical procedures, organ problems, and medications of our patient provide insight to us of the cases that may be encountered, and contributes to necessary surgical-medical guidance and to determine the intensity and type of exercise to be applied.

1q31.3q42.12 trisomy is an extremely rare chromosomal disease. We consider that physical therapy is necessary to prevent possible deterioration and improve the quality of life. We believe our study will guide other cases and contribute to the literature.

Author Contributions

All authors contributed by examining and evaluating the case.

Conflict of Interest

The authors declare that there is no conflict of interest.

Ethical Approval/Informed Consent

Necessary permissions were obtained from ethics committee, with the ethics committee decision no 2020/739 to conduct the study. Necessary information was given to the family and an informed consent form was obtained.

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