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Case Report

Kabuki Syndrome; A rare entity

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ABSTRACT: Kabuki Syndrome is a multiple congenital anomaly/mental retardation syndrome of unknown cause. More than 500 cases of both Asian and non-Asian heritage have now been reported. Although a few associated cytogenic abnormalities have been described, the majority of patients have normal chromosomes and no cause has to date been identified. Diagnosis is based on characteristic dysmorphic features, visceral abnormalities, and clinical features. Ligamentous laxity and joint hyperextensibility have been described in 74-96% of the individuals with this syndrome. Joint dislocation is also present in up to 50 % of individuals with the syndrome. Recurrent patellar dislocation may result in significant disability and require surgical repair joint dislocation may invalue middle ear ossicles and can clead to hearing of imparment. We report a case of 24 years old girl presented with recurrent patellar dislocation and diagnosed as Kabuki Syndrome. Features are discussed and literature reviewedn.
Key Words : Kabuki syndrome, Congenital anomaly, Mental retardation.

INTRODUCTION : Kabuki Syndrome, was first described in Japanese children, in a pair of parallel reports in 1981[1,2] with 20 cases reported in the medical literature by 1985[3]. It is a multiple congenital anomaly/mental retardation syndrome of unknown cause. More than 500 cases of both Asian and non-Asian heritage have now been reported[4]. Although a few associated cytogenic abnormalities have been described[5], the majority of patients have normal chromosomes and no cause has to date been identified. The inheritance pattern of this disorder has not been established. Most cases are sporadic but a few families with multiple generations of affected individuals have been reported, suggesting autosomal inheritance[6]. Diagnosis is based on characteristic dysmorphic features, visceral abnormalities, and clinical features. Ligamentous laxity and joint hyper extensibility have been described in 74-96% of the individuals with Kabuki syndrome[6,7,8]. Joint dislocation is also present in up to 50 % of individuals with the syndrome and is one of the individual features of this syndrome[9]. Recurrent patellar dislocation may result in significant disability and require surgical repair. Dislocation of middle carossicle may occur leading to paracusis and hearing imparirment. We report a diagnosed case of Kabuki Syndrome that presented with recurrent patellar dislocation.

CASE REPORT: A 24-year-old patient was admitted with a painful left knee joint, which according to the patient had “popped out”. She also gave a history of right patellar dislocation a year previously. X-rays showed dislocation of the left patella with no other bony deformities (Fig.1, 2, 3). Past medical history revealed that she developed hearing impairment to low frequencies with bilateral eustachian tube insufficiencies when she was 4 years old, for which she had bilateral grommet insertion and antral washouts. Later on, she developed speech difficulties for which she had speech therapy for a considerable period of time. She also developed contractures of the ring and little fingers of both hands. X rays showed no bony abnormalities apart from flexion of the proximal interphalangeal joints of both hands. She had a global developmental delay and Griffith’s assessment showed 2 years and 1 month when she was 3 years and 9 months. She also exhibited hyper-abduction of both hip joints. General physical examination revealed that she was micrognathic with large ears. Local examination revealed that she had a tender left knee

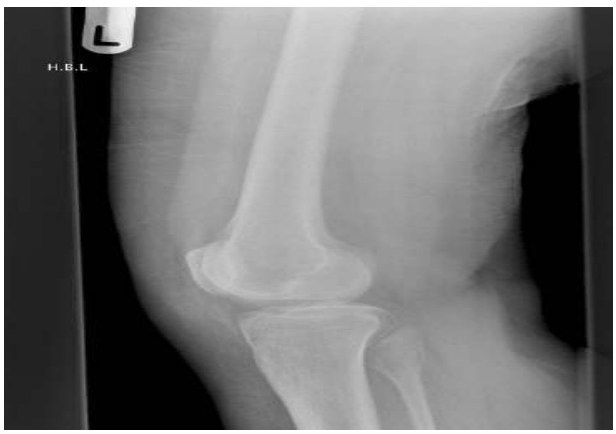

Figure-1 : Pt is diagnosed only on clinical grounds by ENT doctor.

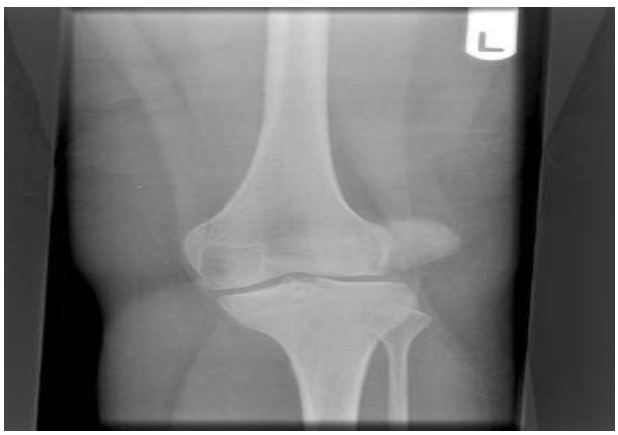

Fig-2 : Dislocation of light patella.

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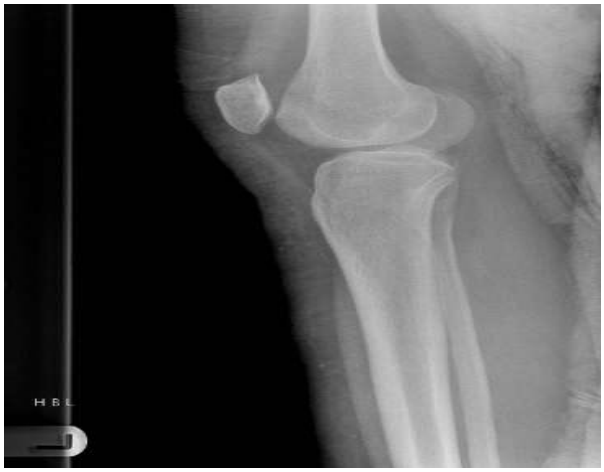

Fig-3 : Patellar dislocation (lotent veiw)

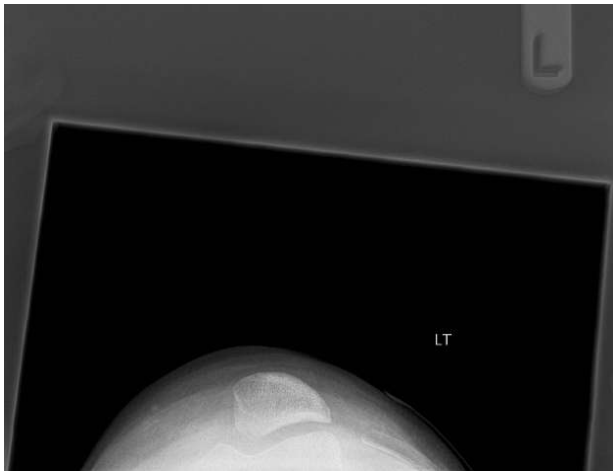

Fig-4 : Re-location under general arareentity.

joint with a laterally displaced and rotated patella. Knee movements were significantly restricted. The SHO on call tried unsuccessfully to reduce it in the accident and emergency department under intravenous sedation. The knee was manipulated under general anesthesia (Fig-4), The patient was sent home the next day and reviewed one week later in fracture clinic where it was noted that she was comfortable and movements of the knee joint were no longer restricted. She was referred to physiotherapy for further rehabilitation.

DISCUSSION : Kabuki Syndrome has encountered in 1967 and reported in 1981[1,2] bears its name because of facial similarities to that of the famous Japanese art form. Though reported initially in Japan, the syndrome has also been reported from South America and Europe. In the last thirty five years since the first description of Kabuki syndrome, over 500 cases in the literature have documented the major and minor malformations associated with this condition[4]. Although multiple authors have included descriptive data of the intellectual performance of patients in their series, no formal analysis of this condition has been under taken. The etiology of the syndrome remains unknown, with familial occurrence estimated at approximately 2%. Few patients have been identified with abnormalities including X chromosome rings[10], translocations /inversions[11], and duplications[12]. The majority of the patients have normal chromosomes and no specific genes to date have been identified. Most of the patients have five cardinal manifestations, namely, the characteristic facial features, skeletal abnormalities, postnatal growth deficiency, mild mental retardation and dermatolyphic abnormalities. The commonly described facial features in this syndrome are arching eyebrows sparse in lateral half, long palpebral fissures, ectropion of lower eyelids, broad nasal tip, high arched palate, mal-occlusion of the teeth and short fifth finger[1,2],.Other features such as low posterior hairline, epicanthal fold, pre-auricular dimple and micrognathia are also seen. Skeletal abnormalities are part of the diagnostic criteria for this condition. The initial reports cited short and incurved 5th digits and short fifth metacarpals, cone shaped epiphysis of the proximal second through fifth phalanges, and various vertebral anomalies (butterfly vertebra, sagittal cleft scoliosis and narrow inter vertebral disc space). Multi organ system has been described, including epilepsy with and without polymicrogyric changes[13,14] hepatic involvement, including fibrosis and sclerosing cholangitis[15], autoimmune disorders, malignancies such as leukaemia and endocrinopathies including diabetes and deficiency of growth hormone. Joint hyper-mobility is seen in one half to three quarters of the patients and joint dislocations are not uncommon, particularly of the hips, patellae and shoulders[8,9]. However it has been found that joint hyper-mobility tends to improve with the age. Patellar dislocation is common among 18% of older children and young adults with this syndrome. The frequency is likely to be affected by reporting bias (overestimate) and an underestimate due to the lack of follow up records in most of the individuals reported. Conductive hearing loss is common in patients with this syndrome and as would be expected, is associated with poor verbal outcomes. Our patient had global developmental delay, developed speech difficulties and had recurrent hearing and upper respiratory infections. Digilio et al reported that 58 % of the patients with this syndrome had congenital heart diseases and considered these cardinal features[16]. In conclusion, patellar dislocation is frequent among older children and young adults with Kabuki Syndrome, especially among female obese individuals with lax knee joints. Research has shown that the sex ratio of individuals with Kabuki syndrome is even, while that of those with the syndrome and patellar dislocation is in favour of females. Although the number of the patients with patellar dislocation and the syndrome is limited, this indicate that being a female may be a risk factor for developing patellar dislocation among the individuals with Kabuki syndrome. The examining doctor should particularly consider this diagnosis in patients who present with distinctive facial features and skeletal deformities. Earlier detection of this syndrome may have important implications for the long-term prognosis of such patient.

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