Abstract. – We report on eight cases of patients affected by KBG syndrome (KBG stands for the initials of the affected patients in the original report), a rare genetic disease, that we find only in 40 cases mentioned in the scientific literature. In this work we present the minimum diagnostic criteria of diagnosis due to identify the syndrome and a hypothesis of study for the research of the involved factors.

Key Words: Macro-olidontia, Short stature, Skeletal anomalies.

Introduction

The KBG syndrome is a rare genetic disease clinically featured by: facies peculiar with macro-olidontia, short stature, mental retardation and skeletal anomalies, associated with microcephaly, deafness, ocular and cardiac anomalies and dermic syndactyli4. In all patients are also present dermatological alterations like thin, opaque and fragile hair and anomalies of fingernails. The most common complicances are: deformity and pain secondary to the skeletal problems, multiple caries and precocious loss of permanent teeth.

Materials and Methods

Eight isolated cases of KBG syndrome (4 males and 4 females within 4 and 12 years old) on a total of 1500 patients monitored between January 1985 and March 2002 in the Center of Medical Genetic Clinic c/o “Moscati” Hospital located in Avellino (Italy).

All the patients have the same features: facies peculiar with low junction of hair either anterior or posterior, wide auricular pavilions and antibacks, bulbous nose, long and flattened filter, advanced thin lip and macroodontia of the maxillary superior incisors (Figure 1). Slight mental retardation and unarticulated walk associated to a mild and sociable behaviour.

Brachidattilya with a PMF (metacarpophalangeal pattern profile) characterized by the shortness of the middle phalanges of the third and the fourth middle finger.

Moreover we noticed specific skeletal anomalies of the column, absence of the XII pairs of ribs, presence of hypoplastic supernumerary cervical ribs (Figure 2), and fusion of some vertebral elements in the second, schisis of the posterior arch of S1 and L5 respectively in the third and in the fourth patient (Figure 3).

We found many variants regarding the dorsal vertebrae with fusion of vertebral elements.

Results

The eight patients were submitted to metabolic screening and to the karyotype exam but the results were negative. In three cases the skeletal age corresponds to the chronological age, in one is delayed and in two of them is superior to the chronological age, we found an advanced skeletal age of 16 years old in the first
patient and in the second patient of 14 years and five months old. In two patients it has been found agenesia and in an other hypoplasia of the frontal sinuses. The ocular anomalies such as megalocornea and squint described in the medical literature were not pointed out but only in a patient it has been diagnosticated a bilateral cataract. In two patients it has been found cryptorchidism. In all the patients it has been noticed low vocal tones.

**Discussion**

The syndrome seems to be not so rare for the some of isolated cases which we have observed. The percentage should be underestimated, especially in the evaluation of the patients that show the minimum signs\(^8\). We must define the minimum criteria of diagnosis: beside to facies peculiar with macrodontia of superior central incisors, beside to the slight mental retardation and the specific anomalies.

The skeletal cord\(^6\) seems to be a constant hit syndrome that has been always underestimated. The other non-specific anomalies of the column such as cifosys and scoliosis and skeletal anomalies (shortness of the palm of the hands and feet) don’t seem to be cataloged in the minimum criteria of this condition. The constant presence of costo-vertebral anomalies\(^10\) in the 8 monitorized cases, such as Hermann’s descriptions, make necessary in this patients radiological evaluation.

The diagnosis of the KBG is predominantly clinical. We tried to demonstrate in an objective way the real mean of the clinical data. For example the macrodontia of the superior central incisors; pantomography shows in our patients the width of the incisors is more than 9.5 mm [normal rate is: among 7.8 mm (3\(^{rd}\)Pc) and 10.4 mm (97\(^{th}\)Pc) in a population of 106 men of race-white; among 6.9 mm (3\(^{rd}\)Pc) and 10 mm (97\(^{th}\)Pc) in a population of 95 women of white race].

We also analyzed the thin, opaque and fragile hair. Starting from the alterations of the hair, gathered with the nails alterations and with the macrodontia and oligodontia and the precocius fall of teeth, seems to be that the KBG is a variant of Ectodermic Dysplasia. Considering all these dermatological alterations, it might be hypothesized that there is more than a factor involved\(^12\). So the syndrome called KBG might be definite such as a syndrome of close factors.
Eight isolated cases of KBG syndrome: a new hypothesis of study

Figure 2. Hypoplastic supernumerary cervical ribs.

Figure 3. Vertebral schisis of L5.
References


