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Congenital Absent Radii without Thumb Aplasia: The First Case in Iraq and the Early Documentation of the Condition in the Literature

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Abstract

Background: The vast majority of the reported cases of congenital absent radii without thumb aplasia were associated with congenital thrombocytopenia or thrombocytopenia occurring early life. The occurrence of congenital absent radii without thumb aplasia and without evidence congenital thrombocytopenia or thrombocytopenia occurring early life is extremely rare and has not been reported in Iraq.

Patients and Methods: An Iraqi newborn infant with congenital absent radii without evidence of congenital thrombocytopenia during the first week is presented, and the relevant literatures were reviewed with aim of describing the early documentation of the condition in the medical literature.

Results: A newborn infant with congenital absence of the radii without thumb aplasia. The neonate was followed for one week without the occurrence of thrombocytopenia. The rest of physical examination was normal and no other abnormalities could be detected. The parents were consanguineous, but family history was negative for a similar condition.

Conclusion: The extremely rare occurrence congenital absence of the radii without thumb aplasia without evidence of thrombocytopenia during the first week of life is reported in an Iraqi newborn.

Keywords: Congenital absence of the radii; Thumb aplasia; Thrombocytopenia

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Introduction

The vast majority of the reported cases of congenital absent radii without thumb aplasia were associated with congenital thrombocytopenia or thrombocytopenia occurring early life. In addition, this condition can be associated with defects of the lower limbs, and anomalies of the gastrointestinal, cardiovascular, and other systems. The association of absent radii with thrombocytopenia was first described in one patient by Greenwald and Sherman [1]. Sixty years later, in 1988 Hedberg from New York City reviewed 100 cases [2,3].

After more than 25 years after the first description of the condition by Greenwald and Sherman [1], Gross et al. [4] reported the disorder in a pair of siblings suggesting an autosomal recessive inheritance. Thereafter, Bell et al. [5] emphasized the peculiar association of bilateral absence of the radii with congenital thrombocytopenia.

In 1957, Emery et al. [6] reviewed 5 cases including the case of Bell et al. [5]. In 1959, Shaw and Oliver [7] reported the birth of an affected sibling to the case the previously described in 1956 confirming an autosomal inheritance of the condition [5].

In 1969, Hall et al. [8] from Baltimore reviewed 40 cases (27 from the literature and 13 new cases) of the condition and emphasized that congenital shortening and deformity of the ulnae may occur in this condition, and the fingers and thumbs are always present. Other skeletal anomalies are frequent.

The occurrence of congenital absent radii without thumb aplasia and without evidence congenital thrombocytopenia or thrombocytopenia occurring early life is extremely rare and has not been reported in Iraq [9,10].

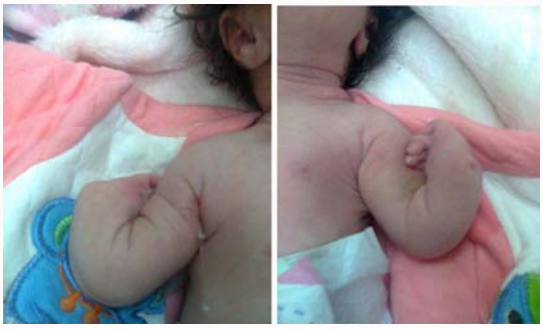

Figure 1: A two-day Iraq neonate with congenital absence of the radii without thumb aplasia.

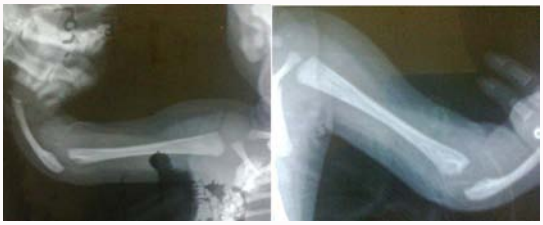

Figure 2: Radiograph showing absence of the radii.

Patients and Methods

An Iraqi newborn infant with congenital absent radii without evidence of congenital thrombocytopenia during the first week is presented, and the relevant literatures were reviewed with aim of describing the early documentation of the condition in the medical literature.

Results

A newborn infant with congenital absence of the radii without thumb aplasia was observed (Figure 1). Absence of the radii was shown on radiograph (Figure 2). The neonate was followed for one week without the occurrence of thrombocytopenia. The rest of physical examination was normal and no other abnormalities could be detected. The parents were consanguineous, but family history was negative for a similar condition.

Discussion

The occurrence of congenital absent radii without thumb aplasia and without evidence congenital thrombocytopenia or thrombocytopenia occurring early life is extremely rare and has not been reported in Iraq [9,10].

Review of the literature revealed that the first well document of case of congenital absence of the radii without thumb aplasia was reported by Secord from Canada [11]. Thereafter during more than one century less than 20 cases have been reported in the literature [12-14].

The patient of Secord was the fifth child of apparently healthy parents. The second child of the parents had a very similar deformity of both arms, and died of inanition at the age of seven weeks. X-rays showed total absence of both radii, without any other congenital deformity. The metacarpal bones are perfect in number and development [11].

Goldenberg reported possibly identical male twins with bilateral complete absence of the radius occurred in association with spina bifida [12].

Conclusion

The extremely rare occurrence congenital absence of the radii without thumb aplasia without evidence of thrombocytopenia during the first week of life is reported in an Iraqi newborn.

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