Case report

Coexistent chromosomal and autosomal dominant conditions in a triple pregnancy

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Introduction

Triplet pregnancy is reported from time to time with various combinations of conditions, such as all three with pyloric stenosis [*1*], all three with hypodontia [*2*], a craniothoracophagus associated with a normally developed infant in uniovular triplets, one of the triplets being an acardiac, acephalic monster [*3*] and dizygous anencephalic triplets [*4*].

Case report

In 1990 at El-Fateh Children's Hospital, Benghazi, triplet babies, two males and one female, were born at term after a normal pregnancy and delivery to a 27 year old normal, healthy mother, who had a two and a half year old healthy girl. The father was 30 years old and in good health. There was no family history of either hereditary or genetic diseases.

Trizygoty was established by clinical evaluation of placentation. Birth weights were 2.6 kg, 1.8 kg and 2.0 kg in order of presentation. The smallest of the triplets was admitted to the nursery from the labour ward because of mild tachypnoea, which resolved after a few hours. He was feeding well, his blood sugar was stable and nothing abnormal was found on clinical examination. He was discharged on day two.

At follow-up two months later, the baby was seen to have multiple brown macules over the trunk and limbs. Ten were observed measuring between 0.5 cm and 1.5 cm in diameter. There was also freckling of the right axilla. The rest of the examination was normal. The first triplet was found to have features of Down syndrome, which was subsequently confirmed as trisomy 21 by cytogenetic studies. The remaining triplet was found to be a normal female.

Discussion

The first of the triplets was Down syndrome with trisomy 21, which had arisen due to nondisjunction (Figure 1). The second child had neurofibromatosis, which is an autosomal dominant condition (Figure 2). Family history revealed that neither the parents nor the grandparents were affected with this condition, and hence this may be a case of spontaneous mutation occurring in the pregnancy

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Figure 1. First triplet with Down syndrome

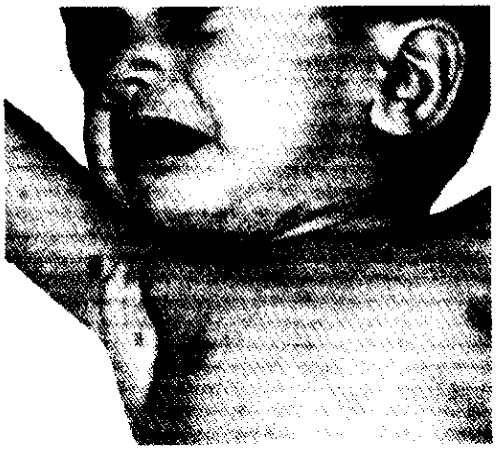

Figure 2. Second triplet with neurofibromatosis

It is rare for two different types of deleterious genetic conditions (chromosomal abnormality and gene mutation) to occur simultaneously but independently in one pregnancy affecting two of the triplets. It is believed that this is the first time that Down syndrome and neurofibromatosis have been reported together.

References

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