CASE PRESENTATION

G Syndrome : A Case Report

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INTRODUCTION

The G syndrome was first reported by Opitz et al. in 1969 in four brothers in one family, G family[1]. Later several other cases were found in the same kindred by Little and Opitz. Since then, to our knowledge, there have been only a few other reported cases of this syndrome[2,3,4,5]. The G syndrome is characterised by distinctive facies with hypertelorism, slanted palpebral fissures, posteriorly oriented external ears, prominent occiput and parietal eminences; hypospadias, with descended testes, is also present. Imperforate anus was present in one of the four original cases. Defective swallowing mechanism with regurgitation and a hoarse cry, wheezing and stridorous respiration have been consistently present.

The purpose of this report is to describe the case of a male infant who had many of the features consistant with the diagnosis of G syndrome. In addition there were certain findings in our case that have not so far been reported in a case of G syndrome (Table 1).

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Table 1
Comparison of features reported in G syndrome with our case (JMA)

Features	*G Syndrome*	*JMA*
SYNDROME (Functional)		
Choking with feeding	+	+
Stridorous breathing	+	+
Wheezing	+	+
Cyanosis	+	+
Weak hoarse cry	+	+
Failure to thrive	+	+
Recurrent aspirations	+	+
SIGNS		
Craniofacial		
1. Prominent occiput	+	+
2. Prominent parietal eminences	+	? ±
3. Hypertelorism	+	+
4. Slanted auricles (Abnormal differentiation)	+	+
5. Slanted palpebral fissures	+	—
6. Puffiness of lower eye lids	—	+
7. Micrognathia	+	+
Genital malformations		
1. Hypospadias	+	+
2. Bifid Scrotum	+	—
3. Hydrocele	—	+
G.I. Tract		
1. Achalasia	±	—
2. Imperforate anus	±	—
3. Short frenum of tongue	±	+
Additional findings noted in JMA		
High-arched palate		+
Partial malrotation of the gut		+
"Eventration" of Diaphragm (Localised)		+

THE CASE

JMA, a 20 day old boy was admitted to Salmaniya Medical Centre with a history of difficulty in breathing and episodes of generalised cyanosis of 2 weeks duration. He was the product of an uneventful full-term pregnancy and normal delivery at home, his birth weight was not recorded. He had 4 sisters and 2 brothers, the parents are not blood related. There was no history of difficulty in swallowing or of wheezing in any of the siblings or parents.

Following admission, physical examination revealed a critically ill infant with generalised cyanosis, distressing cough, severe chest retractions, and he was choking on his copious secretions. His weight was 3030 gms. He had an odd looking facies with marked hypertelorism, prominent occiput, low-set posteriorly slanted ears and a large mouth with a large upper lip. Other abnormal findings present were : a high arch palate, tongue-tie, coronal hypospadias and hydrocele of the right scrotal sac. Both testes were in the scrotum. (Fig. 1, 2, 3).

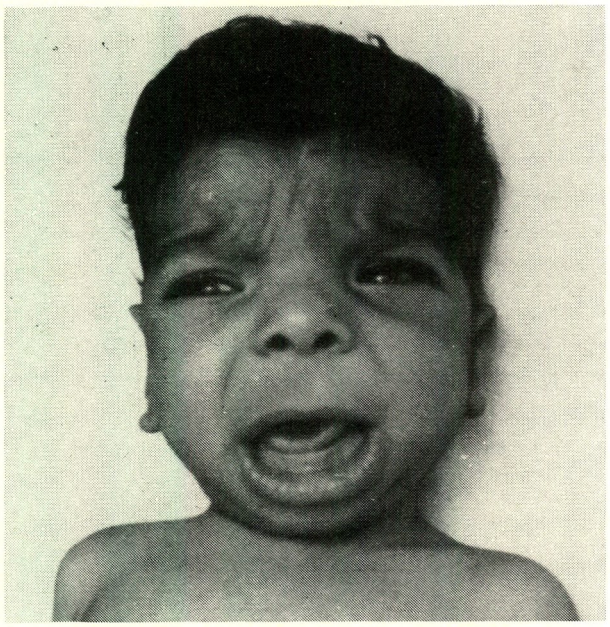


Fig. 1 – Showing the odd looking facies, large mouth, hypertelerism, prominent occiput and a large upper lip.

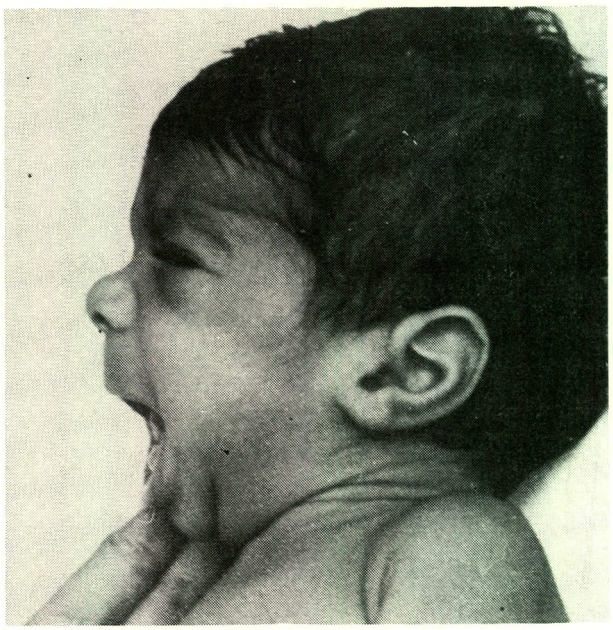


Fig. 2 – Showing the low set posteriorly slanted ears.

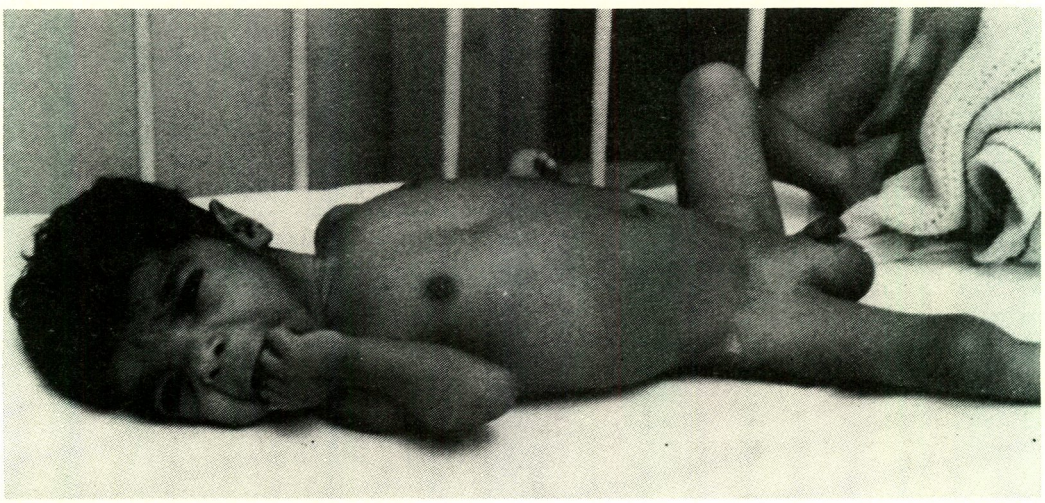


Fig. 3 – Showing the coronal hypospadius and hydrocele.

Bilateral wheezes and rales were audible over both lungs and a chest roentgenogram demonstrated bilateral pulmonary infiltrates, air trapping and atelectasis of the right apical lobe. Also noted was a semicircular, homogenous opacity above the left hemidiaphragm. With improvement in his condition the weak, hoarse cry, wheezing and stridorous breathing became very obvious. He developed repeated episodes of aspiration pneumonitis that were associated with difficulty in swallowing.

Barium swallow examination did not demonstrate either achalasia nor any evidence of tracheo-esophageal fistula. Barium enema ruled out hernia of Bochdalek but it revealed partial malrotation of the gut with the caecum positioned over the vertebral column (Fig. 4). The opacity above the left hemidiaphragm was thought to be localised eventration of the diaphragm (Fig. 5). The patient continued to have a feeding problem but with good nursing care and careful feeding in hospital he did gain weight and developed social smile. He did not appear to have gross mental subnormality. He spent most of his life in hospital and at the age of five and a half months he died suddenly during an attack of severe respiratory distress, probably as a result of aspiration or due to choking on his secretions. His weight at the time of death was 3340 gms. Request for post-mortem was refused by the parents.

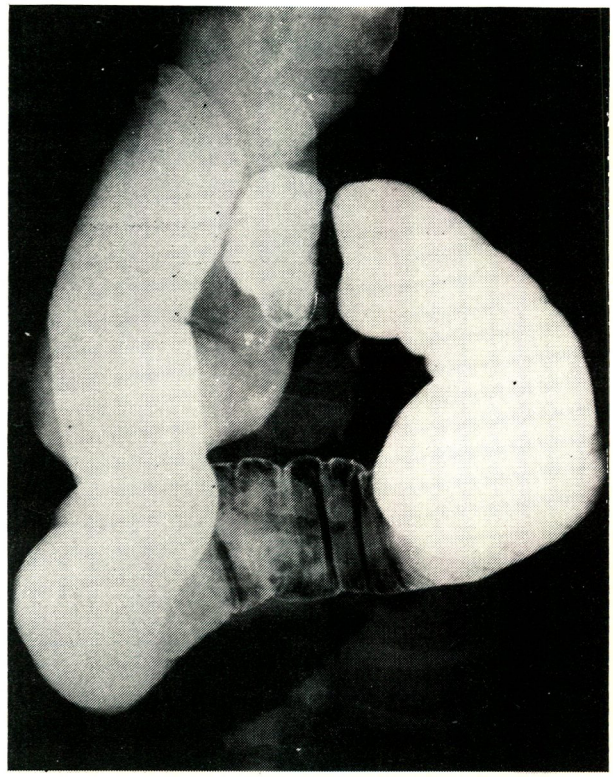
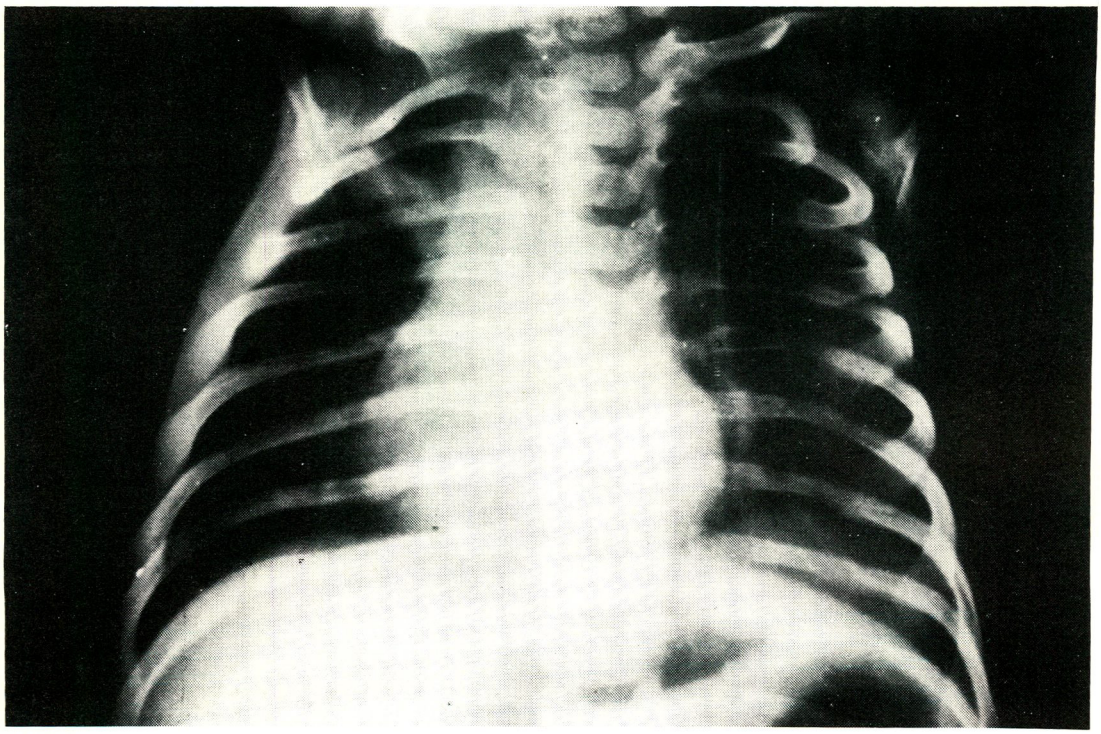


Fig. 4 – Showing partial malrotation of the gut with the caecum positioned over the vertebral column.

Fig. 5 – Showing an opacity above the left hemidiaphragm; localised eventration of the diaphragm.



Blood counts, serum electrolytes, calcium and serum immunoglobin were within normal limits.

DISCUSSION

Many features of this patient are those of a typical case of G syndrome as reported by Opitz et al in 1969. Our patient had in addition certain features which have not so far been reported. These are, partial malrotation of the gut and hydrocele of the right scrotal sac etc. (see Table 1).

Cinefluoroscopic studies of the swallowing mechanism could not be performed. This might have demonstrated the defective swallowing in this patient because his functional difficulties were clearly due to defective swallowing. In at least one of the four original cases of Opitz et al. cine-oesophagram was reportedly normal.

Familial inheritance could not be confirmed in our case. There was no history of swallowing difficulty or of wheezing in any of the siblings or parents. The proposed mode of inheritance of this syndrome is either x-linked (most likely) or autosomal dominant with male sex limitation.

SUMMARY

A case is reported who had many features of G syndrome plus some additional findings, high arched palate, partial rotation of the gut and localised eventration of the diaphragm, none of which have been reported so far in this syndrome.

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