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Lymphocyte studies revealed the mother’s chromosomes to be normal. The father was unavailable for study.

Discussion

The clinical features of patients with deletions of 2p are summarised in the table. The patient of Ferguson-Smith et al is not included in the tabulation because he was also partially trisomic for the distal four bands of 5q and so was not a pure case of partial 2p monosomy. The patient reported by Zachai et al is identical to case 2 of Emanuel et al.1

Given the paucity of reported cases, only the most tentative statements can be made regarding the clinical picture associated with deletions of 2p. The two cases presented by Emanuel et al1 have nearly identical deleted segments and share the following stigmata: mental retardation with microcephaly (−4 to −6 SD), failure to thrive, normal weight and length at birth with postnatal onset of growth deficiency (−4 to −5 SD), delayed closure of the anterior and posterior fontanelles (both patent at 17 months), low set ears, and single flexion crease on the fifth fingers.

Although the two reported cases of interstitial deletions have deleted bands in common (table), the missing segment in the patient of Fryns et al was over three times the length of the deleted segment in the patient of Duca et al,4 making a phenotypic comparison speculative at best. The patients of Fryns et al and Duca et al did, however, show some phenotypic similarities, particularly in the areas of the cranium (frontal bossing with narrow forehead), spine (kyphosis), and toes (long broad big toes with valgus deformity and overlapping toes). Our patient appears to have a single deleted band (2p14) although we cannot rule out the possibility that this band has been incorporated into another breakpoint site. The smallness of the deleted segment may explain her minimal dysmorphogenetic features; however, there is a lack of clinical similarity between our patient and the other two cases of interstitial 2p deletions. For example, our patient had premature closure of her fontanelles, whereas three other patients, including one with a deleted segment incorporating band 2p14,4 had delayed closure of their fontanelles. It is possible that our patient’s physical and mental stigmata are the consequence of a disruption in one or more gene’s nucleotide sequence resulting from this child’s numerous chromosome breaks. Given the uncertainty of our patient’s karyotype and the limited number of 2p deletion cases, it is evident that more cases of 2p deletions are required before a clear cut 2p deletion syndrome, or syndromes, emerges.

References


Correspondence and requests for reprints to Dr Robert S Young, Department of Pediatric Dentistry, University of Texas Health Science Center, San Antonio, Texas 78284, USA.

Sacrococcygeal teratoma and normal alphafetoprotein concentration in amniotic fluid

M SZABÓ*, P VARGA†, A ZALATNAI‡, J HIDVÉGI§, Z TÓTH*, AND Z PAPP*

Department of Obstetrics and Gynaecology, University Medical School, Debrecen*; and Department of Radiology†, Oncopathological Research Institute‡, and 1st Department of Obstetrics and Gynaecology§, Semmelweis University Medical School, Budapest, Hungary.

SUMMARY It is usually assumed that in the case of sacrococcygeal teratoma the concentration of alphafetoprotein in the amniotic fluid is increased. In the case reported here the AFP concentration in the amniotic fluid was found to be normal in spite of a large teratoma not covered with skin. Possible reasons for this are discussed and the histological characteristics of the tumour are reported. It is emphasised that this teratoma could not have been recognised
antenatally by normal AFP screening, but was only possible by ultrasound examination.

Sacrococcygeal teratoma is usually congenital, although in 25% of cases it manifests after the age of 2 months. As its name suggests, this tumour spreads through the perineum and the small pelvis, thus displacing the anus and the external genitalia. It consists of solid and cystic parts, usually surrounded by a connective tissue capsule. It is usually a benign tumour with only 10% of the congenital cases turning out to be malignant according to statistics; however, 90% of cases manifesting after the age of 2 months are malignant.1

This type of tumour acquired new significance with the development of antenatal diagnosis. It was found that in the presence of sacrococcygeal teratoma the AFP concentration and also the acetylcholinesterase (AChE) activity were increased in the amniotic fluid.2-5 More recently, it has been possible to localise the tumour exactly and to determine its size using ultrasound.6

In our case the AFP concentration in the amniotic fluid was found to be normal, in spite of severe sacrococcygeal teratoma. For this reason, we report our antenatal diagnostic and pathological findings.

Case report

Our patient, a 29 year old woman, became pregnant for the first time in the eighth year of her marriage. The serum AFP concentration in the 16th week of gestation was 56 ng/ml. On ultrasound examination (in our region all pregnancies are routinely scanned at 18 weeks), an unusual echo was detected starting in the sacrococcygeal area of the fetus. The lesion was partly solid and partly cystic and measured 53×39×44 mm. It was attached to the pelvis over a considerable area and discontinuity of the spine could be demonstrated posteriorly in the lower lumbar region (figs 1 and 2). Only epithelial cells were found in the amniotic fluid obtained by transabdominal amniocentesis and no cells with phagocytic characteristics indicating an open lesion were observed.7 8 In the amniotic fluid the AFP concentration was 13 230 ng/ml (range 7800 to 19 000 ng/ml) and AChE activity was 9·5 U/l (range 3·3 to 17·8 U/l).

On the basis of these findings we considered the diagnosis of sacrococcygeal teratoma with skin cover and spina bifida. To confirm the diagnosis, ultrasound examination was repeated after two weeks. The tumour showed definite growth (58×42×45 mm) and the closure defect could still be seen.

In view of these findings, we discussed the situation with the couple, who requested termination. Abortion was induced by 100 ml of 0·1% Rivanol solution injected transcervically into the extraovular area and then an oxytocin drip infusion was set up the following day. The abortion took place uneventfully.

The male fetus weighed 350 g. There was a tumour in the sacral and coccygeal area, with a wide...
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The fetus with teratoma after abortion.

FIG 3 The fetus with teratoma after abortion.

base and an hourglass constriction around the middle; the surface was uneven and covered with membrane (fig 3). It was soft to the touch, cyanosed in certain areas, and greyish-yellow in other places. There were also necrosed and decomposed areas in the cut surface. A 15 mm wide open defect was observed at the base of the spine and this area was practically covered with tumour. No other macroscopic pathological alteration was found during the thorough dissection of the fetus.

Hyperplasia of mixed structure and composition could be seen in the histological section of tumour at various places, together with cystic cavities lined with flattened epithelium. The intervening stroma was of myxoid character and distributed at random, usually with mature chondral islets. The greater part of the tumour consisted of embryonic type nerve tissue, arranged into irregular heaps lying close to each other, and with dark, oval nuclei in places, its structure resembling that of retina. The groups of embryonic neural cells were mostly filled with melanin pigment and rosette formation could not be observed. The monomorphic cells formed compact groups, but in certain areas they bulged into a primitive glomerulus lumen. In many places expanded duct-like canals, embryonic hepatic tissue, smooth muscle and striated muscle fascicles, lymphatic capillaries, epithelial cells, and blood forming foci could be seen.

The placenta was both macroscopically and histologically normal.

Discussion

We have found six reported cases of sacrococcygeal teratoma where the AFP concentration of amniotic fluid was high. In four of these cases the AChE activity was also measured and was found to be increased in two cases.2-5 In one published case of sacrococcygeal teratoma, the AFP concentration in the amniotic fluid was not high, but this was at 31 weeks’ gestation.6 In another reported case, at 19 weeks’ gestation neither the amniotic fluid AFP concentration nor the AChE activity was increased.9 Similarly, we observed no increase in either the AFP concentration or in AChE activity in our case.

Since there was a closure defect in the lower spine, the quantitative increase of these chemical components and the appearance of phagocytic cells would have been expected. The absence of chemical and cellular reaction might be related to the fact that the tumour had practically covered the closure defect, thus preventing both transudation and cytological exfoliation. Therefore, spina bifida could only be diagnosed by ultrasound.

Apart from spina bifida, however, in most of the cases reported so far, sacrococcygeal teratoma itself has caused an increase in the amniotic fluid AFP concentration.2-5 The tumour produces protein in large quantities and this has been regarded as the reason for the high amniotic fluid AFP concentration.2 The AFP concentration in serum is also increased in the case of teratomas observed in infancy or in adulthood.10 The AFP produced by the tumour can also enter the amniotic fluid directly when it is not covered with intact skin and therefore the normal AFP concentration of amniotic fluid is not unexpected when intact skin prevents transudation.9

In our case, however, the tumour was not covered with intact skin, only with a thin membrane, so that in theory AFP could have entered the amniotic fluid in large quantities, all the more so since the derivatives of all three germ layers were present in the tumour, including hepatic tissue which is an especially active AFP synthesiser. Though it is feasible that with the advance of gestation the quantity of AFP produced might have increased with the growth of the tumour, by that time it could not have been of use for screening and diagnosis.

This case confirms our routine practice that antenatal screening for congenital malformations can be performed effectively only when AFP and ultrasound are used together. By ultrasound examination we could determine the increase in size of the tumour and identify the spina bifida. On the basis of these objective data we could inform the couple of the serious prognosis of the defects. Because the tumour had been diagnosed before the 20th week of gestation, its size was almost that of the intact parts of the fetus, it was gradually increasing in size, and there was concomitant spina bifida, we were in agreement with the couple when they decided for induced abortion. However, this does not necessarily mean that abortion is the only possible solution in all cases of sacrococcygeal teratoma as there might be cases which can be...
successfully operated upon in the neonatal period. Antenatal examination should be able to define the seriousness of the defect and allow discussion and consideration of the various options in the light of others' experiences. Therefore, such cases should be investigated in centres for antenatal diagnosis and individual cases managed on their own merits, allowing for the particular circumstances of the case.

References

The Nager acrofacial dysostosis syndrome with the tetralogy of Fallot

E THOMPSON*, R CADBURY†, AND M BARAITSER*

*Clinical Genetics Department, The Hospital for Sick Children, Great Ormond Street, London WC1N 3JH; and †Queen Elizabeth Hospital for Children, Hackney Road, London E2 8PS.

SUMMARY A male infant is described with mandibulofacial dysostosis and absent thumbs, consistent with the Nager acrofacial dysostosis syndrome. In addition, the tetralogy of Fallot was present. Major congenital heart malformations occur rarely in this syndrome.

The Nager acrofacial dysostosis (AFD) syndrome, originally described by Nager and de Reynier1 in 1948, is characterised by mandibulofacial dysostosis with radial defects. The facial appearance is similar to the Treacher-Collins syndrome with antimongoloid eye slant, malar and mandibular hypoplasia, and ear hypoplasia. The radial defect involved thumb aplasia or hypoplasia in all cases reviewed by Halal et al2 and was associated with aplasia or hypoplasia of the radius or with radioulnar synostosis in half. The precise mode of inheritance of the Nager syndrome remains unclear, as discussed by Pfeiffer and Stoess.3 There is evidence for autosomal recessive inheritance (reports of affected sibs in two families) and for autosomal dominant inheritance (three families with advanced paternal age suggesting new dominant mutation).

The heart is usually normal in Nager syndrome. Exceptions include reports in which the heart defect is minimal or the case is atypical of Nager syndrome, which we discuss.

We present a clear cut case of Nager syndrome with a congenital heart defect.

Case report

The patient, a male, was the only child of an unrelated healthy Nigerian father and West Indian mother. At the time of conception, the mother was aged 19 years and the father was aged 31 years. He had been undergoing investigations for oligosperma. During the pregnancy the mother had remained healthy apart from vomiting at eight weeks for which metoclopramide was given orally. At 30 weeks' gestation, an ultrasound scan was done and showed polyhydramnios. The baby was born by rapid spontaneous vaginal delivery on the same day. The Apgar score at one minute was 5. The five minute Apgar was 9, following administration of oxygen via a face mask. The gestational age was assessed as 30 weeks by Dubowitz criteria. At birth, the weight was 1070 g (10th centile), the length 40 cm (25th centile), and the head circumference 28 cm (50th centile). The following abnormalities were noted (fig 1): severe micrognathia, malar hypo-
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