Brief Reports

Prenatal Diagnosis of Roberts Syndrome

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Roberts syndrome is a rare autosomal recessive condition with variable phenotype. Severe manifestations are profound tetraphocomelia, cleft lip and palate, hypoplastic nasal alae and oligodactyly with infrequent survival beyond infancy. At least 28 patients from 16 sibships have been reported so far. We recently had the opportunity to monitor the pregnancy at risk for having an affected fetus. This is apparently the first case report of this condition from India.

Case Reports

Case 1: A young non-consanguineous couple was referred to the genetic clinic following the birth of a malformed child who expired after one month. This male child, their first born, was delivered at term after an uneventful pregnancy and labor.

There was no history of exposure to any teratogen. Their description of the anomalies was not satisfactory but they had a photograph of the child *(Fig. 1)*. The striking features on the photograph were a severe mid-facial cleft, hypoplastic nares and tetraphocomelia. This was sufficient to diagnose the child as a case of Roberts syndrome and the couple was counselled re-

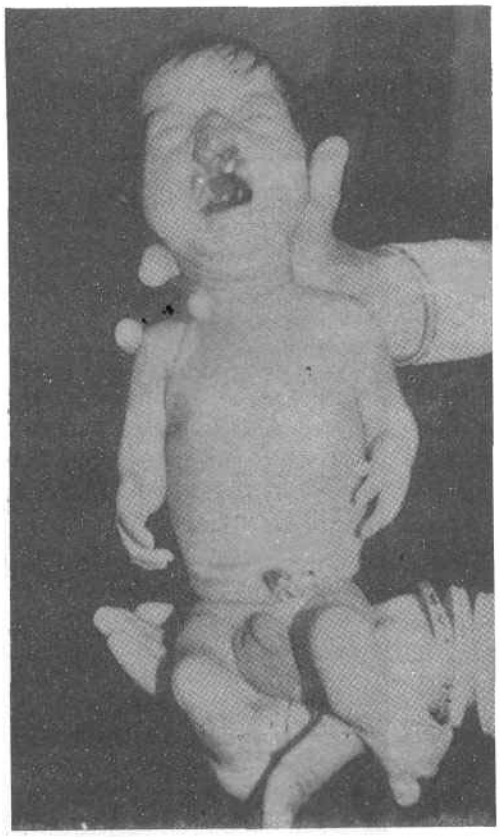

Fig. 1. Clinical photograph of Case 1.

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Received for publication: August 14, 1993; Accepted: June 2, 1994

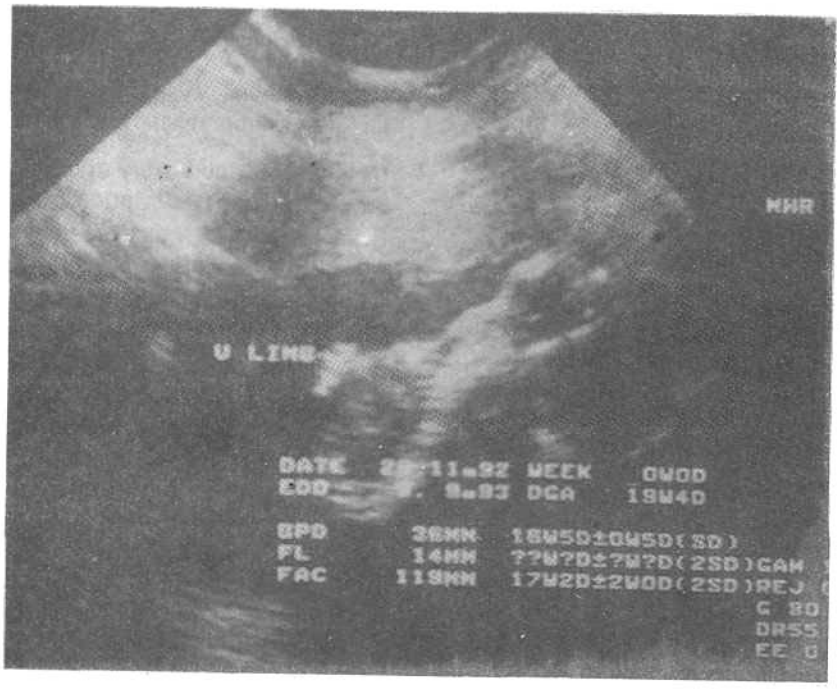

Fig. 2a. USG picture showing short upper limbs.

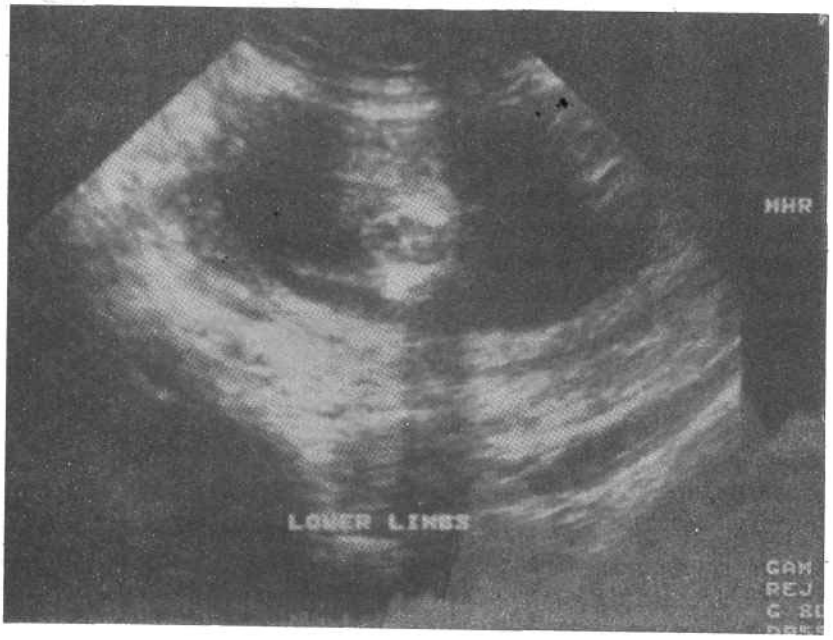

Fig. 2b. USG picture showing short femora

possibility of prenatal diagnosis by ultrasound.

Case 2: The mother's subsequent pregnancy, 1 year later, was monitored by ultrasound. At 16 weeks gestation ultrasonography demonstrated short upper and lower limbs *(Figs. 2a & 2b)*. Femur length was 14 mm (corresponding to 12 weeks gestation). The amniotic fluid was also diminished. On the basis of these findings the pregnancy was terminated at 19 weeks resulting in a still born male fetus *(Fig. 3)*.

The fetus weighed 160 g; measured 18 cm and had a head circumference of 14.5 cm. Limb abnormalities included tetraphocomelia and oligodactyly, *i.e.,* 4 digits on each upper limb. The most notable facial abnormalities were major degree of facial cleft, nasal hypoplasia, hypertelorism, protuberant eyes and mild micrognathia. Roentgenograms further demonstrated shortness of the long bones. Autopsy was performed but no additional abnormalities were found. Autopsy tissue cultures failed to yield fibroblasts suitable for chromosomal analysis.

Fig. 3. Clinical photograph of Case 2.

Discussion

We have described two sibs with a rare autosomal recessive disorder and demonstrated that photographic record is a useful tool for accurate diagnosis, prediction of recurrence risk and also prenatal diagnosis.

Prenatal diagnosis of this condition has been reported infrequently, once by plain roentgenogram during late pregnancy(2) and on two occasions by a second trimester ultrasonography(3,4). Shortness of limbs is simple to detect on a scan and is present in all cases of this syndrome; whereas facial clefting is difficult to diagnose specially in cases of oligohydramnios and may not always be present.

Another diagnostic feature is cytogenetic evidence of abnormal centromere and chromatid apposition (ACCA) or "puffing apart" in heterochromatic and centromeric regions. Unfortunately, we were unable to demonstrate this phenomenon.

In pregnancies considered to be at risk, an early mid trimester scan (at 16 weeks) can prove diagnostic as in our case. Cordocentesis and demonstration of the chromosomal abnormality from fetal blood can also be carried out at the same time.

REFERENCES

1. Wertelecki W. *In:* Birth Defects Encyclopedia. Ed Buyse ML. Oxford, Blackwell Scientific Publications, 1991, p 1498.
2. Freeman MVR, Williams DW, Schimke RN, Temrtamy SA, Vachier E, German J. The Roberts syndrome. Clin Genet 1974, 5: 1-16.
3. Kaffe S, Rose JS, Godmilow L, *et al.* Prenatal diagnosis of renal anomalies. Am J Med Genet 1977,1: 241-251.
4. Robins DB, Ladda RL, Thieme GA, Boal DK, Emaneel BS, Zackai EH. Prenatal detection of Roberts-SC phocomelia syndrome. Am J Med Genet 1989, 32: 390-394.

Congenital Pneumonia Caused by *Klebsiella pneumoniae*

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Pneumonia presenting at birth or occurring within first 48 hours of life is most commonly caused by *E. coli, Streptococci, Klebsiella aerobacter* or *Enterococci(1).* Congenital pneumonia produces intrauterine death in 16-20% of neonates(l). Early diagnosis and adequate treatment can reduce the mortality to a great extent. We report a case of congenital pneumonia caused by *Klebsiella pneumoniae* with successful outcome.

A three hour old, fullterm neonate delivered vaginally weighing 2.36 kg, presented with severe respiratory distress from time of birth. There was no history suggestive of birth asphyxia. Antenatally, the mother had irregular fever for 15 days prior to child birth. On examination, the baby was in severe respiratory distress with respiratory rate of 176 per minute with severe indrawing of chest wall. There were no auscultatory findings in the respiratory system. The chest X-ray showed bilateral infiltrates suggestive of pneumonia (*Fig. 1).* A diagnosis of congenital pneumonia was made.

Investigations showed a hemoglobin level of 12 g/dl, total leukocyte count 6,400 cells/cu mm with 74% neutrophils and 25% lymphocytes, and ESR 22 mm/hr. The absolute band cell count was 650 and toxic granulations were seen in the peripheral smear. Platelets were adequate. The blood culture of the baby and mother's vaginal swab culture were sterile. Culture of the throat swab yielded *Klebsiella pneumoniae* sensitive to cefotaxime and amikacin. The child was treated with parenteral cefotaxime in the dose of 125 mg twice daily and amikacin 20 mg twice daily for 14 days.

A steady decrease in respiratory rate was noticed after 2 days and by the 4th day, the child was able to suck at the breast. On the 14th day of treatment, a repeat chest **X-**

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Received for publication: August 12, 1993; Accepted: June 2, 1994,