On 24.6.83, a 24 year old primipara in good health gave birth to a stillborn male fetus with multiple defects and the appearance of Potter's syndrome. Birth weight was 1775 g after a gestation of 36 weeks and a pregnancy complicated by polyhydramnios. At necropsy there was bilateral renal agenesis, absent ureters (bladder present), absent penis, imperforate anus, and a persistent truncus arteriosus. Owing to collapse of the skull, the cranium was not opened.

Following this, in November 1983, a four month macerated fetus required induction and delivery by D and C, and ten months later she delivered a missed abortion.

Because of this early fetal loss in conjunction with the previous malformed infant a karyotype was done on her and her husband. She showed a female karyotype with two apparently balanced translocations: 46,XX,t(2;13)(p21;q32) and t(5;14)(q33;q32). This indicated a very 'high' risk of reproductive failure. Her mother had one of the balanced translocations and had had two spontaneous abortions, as well as two sons and this daughter. One son, whose wife is currently 13 weeks pregnant, was found to have a normal karyotype. The family history showed a maternal female cousin who had Down's syndrome with no karyotype available.

This case demonstrates that recurrence risks for perinatally lethal renal disease in association with multiple defects may not only relate to autosomal recessive genes, as suggested by Bankier *et al*, but may also show a chromosomal cause. It is therefore important to karyotype such infants because of the consequent high risk implications to other family members.

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Mosaic trisomy 9

SIR,

In 1982¹ we published a case of mosaic trisomy 9 and delineated the clinical phenotype of this unusual trisomy. As there have been fewer than 30 patients reported so far, and since many of them died during the first year of life, there is little information about their development. For this reason we wish to report the development of our patient, who is now 4½ years old.

At 6 months the patient, a girl, had a urinary infection. Pyelography showed mild left pelvic dilatation. Cardiological studies were normal. At 20 months she had a convulsive episode; as she has been taking luminal ever since, the episode did not recur. When she was 2 years she was operated on for cataracts in the right eye. She has dorsal kyphoscoliosis and pes valgus.

With regard to developmental milestones, she stood up at 22 months and walked at 4 years. At present she does not speak. She is severely mentally retarded and has severe microcephaly (45 cm=50th centile for 1 year). Her mother became pregnant again last year and prenatal diagnosis showed a 46,XX karyotype.

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Reference

- 1 Sánchez JM, Fijtman N, Migliorini AM. Report of a new case and clinical delineation of mosaic trisomy 9 syndrome. *J Med Genet* 1982;19:384-7.

Spondyloepiphyseal dysplasia tarda with progressive arthropathy: a rare disorder frequently diagnosed among Arabs

SIR,

Spondyloepiphyseal dysplasia tarda with progressive arthropathy (SEDPA) is a newly delineated clinical disorder,^{1,2} with strong evidence of autosomal recessive inheritance.^{1,3} The disorder bears a striking resemblance to rheumatoid arthritis but differs radiologically owing to the absence of destructive bone changes and the presence of dysplastic bone changes in addition to the platyspondyly. The lack of response to anti-rheumatoid treatment is also a distinguishing feature.

We recently examined a 30 year old Arab female patient with double first cousin Lebanese parents. They had six normal daughters and two mid-trimester miscarriages; there was no history of a similar condition in the family.

She appeared normal for the first 2½ years of life after which she developed difficulty in walking, swelling of the knees and small joints of the hands, followed by gradual stiffness in the knees. At the age of 5 years, a right genu valgum deformity was corrected surgically. Arthropathy of almost all limb joints was progressive and involved the elbows, wrists, hips, knees, and metaphalangeal and interphalangeal joints. By the age of 10 years, the small