

Fetal megacystis at 10–14 weeks of gestation

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Key words: MEGACYSTIS, CROWN-RUMP LENGTH, BLADDER DIAMETER

ABSTRACT

During the study period, 24 492 pregnant women attended the Harris Birthright Research Centre at 10–14 weeks of gestation, at which time, in addition to the measurements of nuchal translucency thickness and crown–rump length (CRL), data on fetal abnormalities were recorded onto a computer database. Cases of megacystis were identified and the records were reviewed. Additionally, the relationship of the longitudinal bladder diameter with the CRL and the bladder diameter/CRL ratio (expressed as a percentage) were examined with the use of data from 300 normal fetuses at 10–14 weeks.

Megacystis was present in 15 of the 24 492 pregnancies (1 in 1633) and in these cases the minimum longitudinal bladder diameter was 8 mm and the minimum bladder diameter/CRL ratio was 13%. In the 300 control fetuses the bladder was visualized in 278 (92.7%) of the cases and the longitudinal bladder diameter increased with the CRL (bladder diameter = $0.065 \times \text{CRL} - 0.69$; $r = 0.47$, $p < 0.001$), none of the measurements was more than 6 mm and the median bladder diameter/CRL ratio was 5.4% (range 0–10.4%) which did not change significantly with gestation ($r = 0.1$, $p = 0.09$). The bladder was visible in all cases with a minimum CRL of 67 mm. In three of the 15 cases with megacystis, there were chromosomal abnormalities. In the chromosomally normal group, there were seven cases with spontaneous resolution, whereas in four cases there was progression to severe obstructive uropathy.

The bladder diameter was 8–12 mm and the bladder diameter/CRL ratio 13–22% in all cases with resolution and in one case with progressive megacystis; in the other three cases with progressive obstruction, the bladder length was more than 16 mm and the bladder diameter/CRL ratio was more than 28%.

INTRODUCTION

The prenatal diagnosis of megacystis and varying degrees of hydronephrosis and hydronephrosis during the second trimester of pregnancy, as well as the association with chromosomal defects and renal damage, have been extensively reported during the last 20 years. However, no

ultrasonographic screening studies have been reported on the prevalence of obstructive uropathy and the natural history of this condition. This study examines the prevalence and consequences of fetal megacystis diagnosed at 10–14 weeks of gestation.

METHODS

Since September 1992 an ultrasound screening study has been implemented at the Harris Birthright Research Centre for Fetal Medicine involving measurement of fetal nuchal translucency thickness at 10–14 weeks of gestation¹. In addition to the measurements of translucency thickness and crown–rump length (CRL), data on any obvious fetal abnormalities were recorded onto a computer database at the time of the ultrasound examination. Details on pregnancy outcome were obtained from the patients themselves or their hospitals. One of the abnormalities searched for was megacystis (Figure 1), and in such cases the longitudinal diameter of the bladder was recorded and follow-up scans were undertaken.

A computer search of the database was made to identify all pregnancies with live fetuses at the 10–14-week scan and an estimated date of delivery before June 1996. In all cases of megacystis the records were reviewed and in the live births the mother and general practitioners were contacted for details on the condition of the child.

The relationship between the longitudinal diameter of the fetal bladder with the CRL and the bladder diameter/CRL ratio (expressed as a percentage) were examined by regression analysis from the study of 300 consecutively examined fetuses at 10–14 weeks of gestation.

RESULTS

During the study period there were 24 492 singleton pregnancies with live fetuses at 10–14 weeks of gestation and the diagnosis of megacystis was made in 15 (1 in 1633 or 0.06%) of the cases. In these 15 cases the minimum

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longitudinal diameter of the fetal bladder was 8 mm (Figure 2), and the minimum bladder diameter/CRL ratio was 13% (Table 1).

In the 3000 control fetuses the bladder was visualized in 278 (92.7%) of the cases and the longitudinal bladder diameter increased with the CRL (bladder diameter = $0.065 \times \text{CRL} - 0.69$; $r = 0.47$, $p < 0.001$), but within the gestational age range that was examined none of the measurements was more than 6 mm. In the control group,

the median bladder diameter/CRL ratio was 5.4% (range 0–10.4%) and this ratio did not change significantly with gestation ($r = 0.1$, $p = 0.09$). The bladder was visible in all 65 cases with a minimum CRL of 67 mm (corresponding to 12 weeks and 5 days of gestation), but this was not seen in 22 of the 235 (9.4%) with a CRL of 38–67 mm (Figure 3).

In three of the 15 cases there were chromosomal abnormalities, with one case each of trisomy 21, trisomy 13 and



Figure 1 Megacystis in a fetus at 12 weeks of gestation

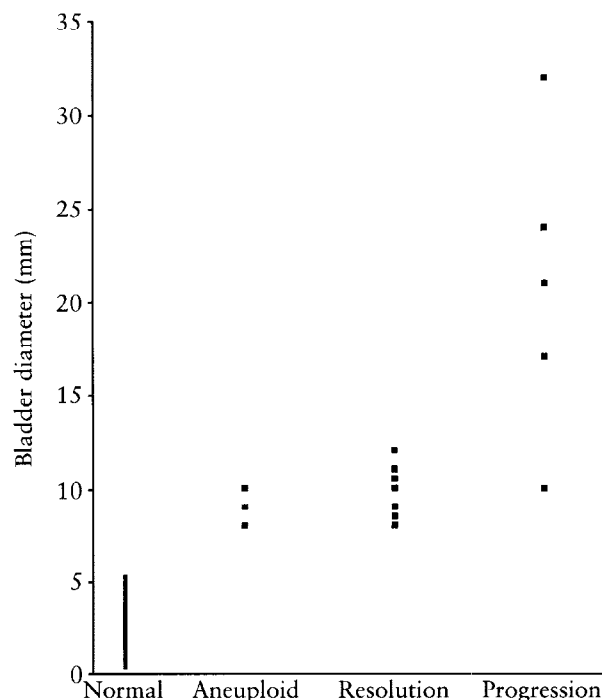


Figure 2 Longitudinal bladder diameter in the 15 cases with megacystis – three chromosomally abnormal fetuses (aneuploid), five cases with evidence of obstructive uropathy ending in fetal death (progression) and seven cases in which the megacystis resolved (resolution). The vertical line represents the range of measurements in 300 normal fetuses

Table 1 Gestation at presentation, crown–rump length (CRL), longitudinal bladder diameter, bladder diameter as a percentage of CRL, fetal nuchal translucency thickness (NT), fetal karyotype, results of 16–20-week anomaly scan and pregnancy outcome in 15 pregnancies complicated by fetal megacystis at 10–14 weeks of gestation

Case	Gestation (weeks)	CRL (mm)	Bladder diameter (mm)	Bladder diameter/CRL (%)	NT (mm)	Karyotype	Megacystis at 16–20 weeks	Outcome
1	12 + 0	57	10	17.5	3.2	46XY + 21	—	termination 13 weeks
2	11 + 6	55	9	16.4	3.8	46XY + 13	—	termination 12 weeks
3	11 + 0	44	8	18.2	1.3	46XY u14/20	resolved	termination 18 weeks
4	12 + 6	54	8	14.8	1.4	46XY	resolved	live birth 41 weeks
5	10 + 1	45	8	17.8	3.0	46XY	resolved	live birth 38 weeks
6	11 + 6	55	10	18.2	2.2	46XY	resolved	live birth 36 weeks
7	13 + 0	70	9	12.9	1.8	46XY	resolved*	live birth 41 weeks
8	13 + 2	74	10	13.5	3.1	46XY	resolved*	live birth 40 weeks
9	12 + 4	62	11	17.7	3.0	46XY	resolved*	live birth 41 weeks
10	11 + 6	55	12	21.8	2.0	46XY	resolved†	live birth 40 weeks
11	12 + 0	56	10	17.9	1.7	46XY	increased*	termination 16 weeks
12	11 + 5	53	24	45.3	2.0	46XY	increased*	termination 14 weeks
13	12 + 3	61	17	27.9	2.0	46XY	shunting*	miscarriage 15 weeks
14	13 + 0	69	32	46.4	3.0	46XY	shunting*	miscarriage 14 weeks
15	11 + 5	52	21	40.4	1.8	—	—	miscarriage 12 weeks

*, Hydronephrosis; †, hydronephrosis at birth

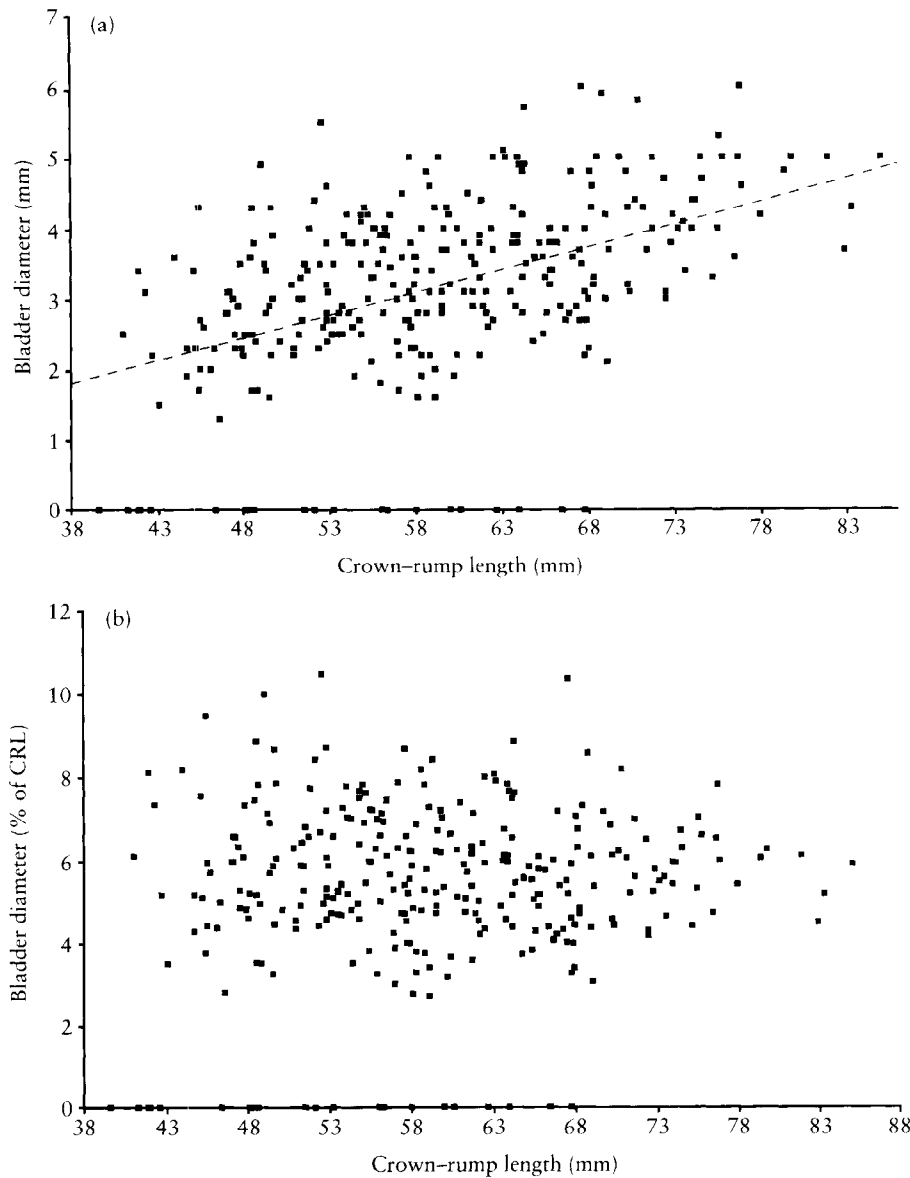


Figure 3 Longitudinal fetal bladder diameter (a) and bladder diameter as a percentage of fetal crown-rump length (CRL) (b) in relation to CRL in 300 consecutive normal fetuses at 10–14 weeks of gestation

an unbalanced translocation involving chromosomes 14 and 20. In 11 cases the fetal karyotype was normal, or phenotypically normal babies were born. In one case there was spontaneous abortion and the fetal karyotype is not known. The fetal nuchal translucency thickness was above the 95th centile of the normal range for CRL in six (40%) of the fetuses with megacystis, including both of the cases of fetal trisomy.

In the chromosomally normal group there were seven cases in which there was spontaneous resolution of the megacystis by 20 weeks of gestation. Although in four of these cases there was pyelectasia at 20 weeks, postnatally only one had mild hydronephrosis and in all other cases both kidneys were normal.

In four cases follow-up scans demonstrated enlargement of the megacystis. In one case termination of pregnancy was carried out at the request of the parents and post-mortem examination demonstrated megacystis and dysplastic

kidneys. In two cases vesicoamniotic shunting was carried out at 14 weeks of gestation, but in both cases there was spontaneous abortion within 1 week of the procedure. In another case, at the 16-week scan there was bilateral hydronephrosis with echogenic renal cortices; analysis of fetal urine obtained by urodochocentesis demonstrated high urinary calcium concentration (2.1 mmol/l)² and the parents opted for termination of the pregnancy.

At the 10–14-week scan, the fetal bladder length was 8–12 mm and the bladder diameter/CRL ratio was 13–22% in all seven cases with spontaneous resolution of the megacystis, in the three cases with chromosomal abnormalities and in one of the four cases with progressive megacystis; in the other three cases the bladder length was more than 16 mm and the bladder diameter/CRL ratio was more than 28%. The bladder was also very enlarged in the one case of spontaneous miscarriage (bladder diameter 21 mm).

DISCUSSION

This ultrasound screening study has demonstrated that at 10–14 weeks of gestation the prevalence of fetal megacystis is about 1 in 1600 pregnancies. In half of the cases there was spontaneous resolution of the megacystis without any obvious adverse consequences on the development of the urinary system or renal function. However, in some cases the megacystis was associated with chromosomal defects and in others there was progressive obstructive uropathy.

In the normal controls, there was a significant increase in bladder length with CRL, but at 10–14 weeks of gestation the longitudinal diameter of the fetal bladder was always less than 6 mm and the bladder diameter/CRL ratio was less than 10%. The fetal bladder was always visualized if the CRL was more than 67 mm, but not in 9% of those with a CRL of 38–67 mm. These findings are similar to those of Green and Hobbins³, who used transabdominal ultrasound to examine 143 pregnancies at 10–13 weeks of gestation and were able to visualize the bladder in all cases by 13 weeks of gestation, compared to only 50% at 10 weeks. More recently, Braithwaite and colleagues⁴ examined 264 pregnancies at 12–13 weeks of gestation and reported that, with the use of both transabdominal and transvaginal sonography, the fetal bladder was visualized in 98% of the cases.

During the second trimester of pregnancy, there is a well-recognized association between renal abnormalities and a wide range of fetal chromosomal defects⁵. The findings of the present series suggest that there may also be an association between chromosomal defects and megacystis at 10–14 weeks. Furthermore, the data suggest that, as with other fetal abnormalities such as exomphalos⁶ and ventriculomegaly⁶, chromosomal defects are more commonly associated with the milder degrees of the given abnormality. In both of our trisomic fetuses, in addition to the megacystis there was increased fetal nuchal translucency thickness, and therefore, the independent contribution of megacystis to the risk for chromosomal defects remains to be determined by larger series.

Severe megacystis at 10–14 weeks (minimum longitudinal bladder diameter of 17 mm) was associated with progressive obstructive uropathy in all cases. In contrast, in the majority of chromosomally normal fetuses with mild or moderate enlargement of the bladder (8–12 mm), there was spontaneous resolution of the megacystis by 20 weeks of gestation without any obvious adverse effects on renal development and function. However, in one of our eight chromosomally normal cases with mild or moderate megacystis there was progression to severe obstructive uropathy. These findings could provide the basis for the development of guidelines in counselling of parents as to the likely evolution of the megacystis.

Extensive animal studies have demonstrated that obstructive uropathy causes renal dysplasia and that the degree of renal damage is related both to the onset and to the duration of the obstruction^{7,8}. Furthermore, such studies have shown that renal damage can be reduced by intrauterine surgery to bypass the obstruction^{8,9}. However, the data from vesicoamniotic shunting in human fetuses

with obstructive uropathy have not provided conclusive evidence that such interventions are beneficial, possibly because by mid-gestation, when surgery is usually undertaken, irreversible renal damage may already have occurred. As suggested from animal studies, first-trimester diagnosis of megacystis and vesicoamniotic shunting could potentially prevent the subsequent development of renal damage. However, as demonstrated in two of our cases, such early interventions could be associated with a high risk of miscarriage, which would outweigh any potential benefits.

This study has established a reference range of fetal bladder size at 10–14 weeks of gestation and has demonstrated that, in the majority of cases of mild-to-moderate megacystis, there is spontaneous resolution. However, this is not always the case and therefore further assessment with serial ultrasound examinations is necessary. In contrast, severe megacystis is likely to evolve into severe second-trimester oligohydramnios and renal dysplasia. An additional finding of the study is the possible association between mild-to-moderate megacystis and chromosomal defects, but the true significance of this finding requires further investigation.

ACKNOWLEDGEMENT

This study was supported by a grant from the Fetal Medicine Foundation (registered charity no. 1037116).

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