PERINATOLOGY

Prenatal Diagnosis of Renal Disease

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Introduction. **The purpose of this study is to report the range of renal congenital anomalies identified by ultrasonography and to analyze the indications for the ultrasound study that lead to their diagnosis.**

Materials and Methods. **All cases of renal malformations diagnosed at our institution from June 2001 through May 2004 were evaluated retrospectively. The indications for sonographic evaluation were reviewed. Cases were divided into those referred for routine ultrasound screening and those referred with other indications. Results were expressed as percents of total.**

Results. **A total of 117 cases of renal congenital anomalies were identified, in 14 cases (11.9%), other congenital anomalies were also present. Hydronephrosis was the most common diagnosis detected in 64 cases (54.7%). There were 21 lethal renal anomalies identified (17.9%). Multicystic dysplastic kidneys were present in 21 cases (17.9%). Renal agenesis was identified in 15 cases (12.8%). Other detected anomalies were: bladder outlet obstruction 15 (12.8%), echogenic kidneys 13 (11.1%), ectopic ureteral implantation 2 (1.7%), renal cysts 1 (0.8%), pelvic kidney 3 (2.6%), double collecting system 1 (0.8%), and unilateral atrophic kidney 1 (0.8%). A total of 94 cases (80.3%) had no indications for sonographic evaluation other than routine screening.**

Conclusions. **Relying on risk factors as indications for ultrasound studies will not detect the majority of congenital anomalies, especially those associated to the urogenital system, a group of conditions that may particularly benefit form prenatal detection.**

Key words: Renal anomalies, Ultrasound, Prenatal diagnosis, Hydronephrosis, Indications.

Prenatal diagnosis of congenital anomalies is one of the most challenging and exciting topics in perinatology. Since many birth defects occur sporadically in patients without identifiable risk factors, exclusive screening of high risk populations fails to identify a large number of cases. In recent years, obstetric ultrasound has become increasingly available. Most practitioners now rely on ultrasound for the prenatal diagnosis of congenital anomalies and fetal evaluation. With improvements in technology, and more pregnant women undergoing these procedures, there has been enhanced diagnosis of congenital malformations (1).

The success in the detection of congenital anomalies depends greatly on the specific anomaly suspected and its prevalence in a particular population, as well as the gestational age at which the evaluation is performed. For example, prenatal diagnosis of gastrointestinal and cardiac anomalies have sensitivities as low as 16 and 23% respectively (2,3). However, renal anomalies, especially those associated with obstruction, have higher detection rates (4).

The purpose of this study is to analyze the incidence range of fetal renal anomalies in a mixed population at a tertiary care facility and to evaluate the indications for referral.

Materials and Methods

This descriptive study was conducted at the University Hospital, a tertiary care center affiliated to the University of Puerto Rico School of Medicine. The majority of patients referred to our institution come from general OB/Gyn or primary practitioners located in different areas of this Caribbean island with a population of approximately 4 million inhabitants, mostly Hispanics. The data was analyzed retrospectively. All examinations were performed by one of two physicians with extensive experience in ultrasongraphy using either a General Electrics Series Logic 500 (General Electric, Milwaukee, WI, USA) or an ATL HDI 1000 (Advanced Technology Laboratories, Seattle, WA, USA) ultrasound.

We defined hydronephrosis as an antero-posterior renal pelvis diameter greater than 1.0 cm. measured in a

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transverse plane in conjunction with calyceal dilatation. All cases meeting these diagnostic criteria were included in the analysis. Cases without calyceal dilatation or with a diameter of 1.0 cm or less were labeled as pyelectasis and excluded from the study.

Maternal age, parity, associated risk factors, diagnosis, and the presence of other non-renal anomalies were evaluated and the indications for sonographic evaluation were reviewed. The cases were divided into two groups, based on the indication for referral. The group of patients referred for routine screening (routine ultrasound) or the group of patients referred with other indications such as an abnormal maternal serum screen, advanced maternal age (age ≥ 35 years old at time of delivery), maternal drug use or exposure, and a family history of congenital anomalies. Results were tabulated and expressed as percents of total. All procedures in this study were in accordance with the ethical standards of our institution and the Helsinki Declaration of 1975, as revised in 1983.

Results

We performed 7,714 sonographic evaluations from June 2001 to May 2004 and identified a total of 117 cases of congenital renal anomalies. One out of every 60.8 sonograms performed at our evaluation unit identified the presence of a congenital renal anomaly. Table 1 summarizes the characteristics of patients in which a renal anomaly was identified.

Table 1. Population Characteristics in Cases with a Prenatal Diagnosis of Renal Disease. (N = 117)

Characteristics	Mean	Range
Maternal age	26.4	15-43
Gravity	2.2	1-9
Gestational age at diagnosis	27.4	12-38
Associated non-renal anomalies	14(12%)	
Male/female ratio	1.7:1	

Among patients with renal anomalies, the average maternal age was 26.4 years (range 15-43). The average parity was 2.2 (range 0-9). In 14 cases (11.9%), other congenital anomalies were also present and the male to female ratio was 1.7:1.

The range of renal anomalies identified is summarized in Table 2. Hydronephrosis (either bilateral or unilateral) was the most common renal defect seen (64 cases) for a 54.7% incidence. There were 21 lethal renal anomalies (17.9%), multicystic dysplastic kidneys (either unilateral or bilateral) were present in 21 cases (17.9%) and renal agenesis (either unilateral or bilateral) was identified in 15 cases (12.8%). Other detected anomalies were: bladder outlet obstruction 15 (12.8%), echogenic kidneys 13 (11.1%), ectopic ureteral implantation 2 (1.7%), renal cysts 1 (0.8%), pelvic kidney 3 (2.6%), double collecting system 1 (0.8%), and unilateral atrophic kidney 1 (0.8%).

Table 2. Range of Prenatally Diagnosed Renal Diseases (N=117)

Diagnosis	# of cases	% of total
Hydronephrosis	64	54.7
Unilateral	43	36.8
Bilateral	21	18
Multicystic dysplastic kidney	21	17.9
Unilateral	18	15.4
Bilateral	3	2.5
Renal agenesis	15	12.8
Unilateral	8	6.8
Bilateral	7	6
Echogenic kidneys	13	11.1
Bladder outlet obstruction	15	12.8
Ectopic ureteral implantation	2	1.7
Pelvic kidney	3	2.6
Renal cysts	1	0.8
Double collecting system	1	0.8
Atrophic kidney	1	0.8

The distribution of cases based on indications for sonographic evaluation is presented in Table 3. A total of 94 cases (80.3%) had no risk factors or indications for sonographic evaluation. They were ordered routinely for the purpose of screening to rule out congenital anomalies.

Table 3. Indications for Sonographic Evaluation Leading to a Diagnosis of Renal Disease

Indication for study	# of cases	% of total
Screening in low risk patient	94	80.3
Abnormal serum screening	5	4.3
Maternal disease	5	4.3
Advanced maternal age (>34)	11	9.4
Maternal drug use	2	1.7
Bleeding during pregnancy	1	0.8
Suspected growth restriction	2	1.7
Previous child with a congenital anomaly	1	0.8

The other 23 detected cases (19.7%) were the product of sonographic evaluations ordered based on several indications including: results of an abnormal serum screen (5), maternal systemic diseases such as diabetes mellitus or chronic arterial hypertension (5), advanced maternal age (11), maternal drug use (2), a suspicion of intrauterine growth restriction based on the identification of a uterine size smaller than those expected for gestational age (2), bleeding during pregnancy (1), or a history of having a previous child born with a congenital anomaly (1).

Discussion

Routine sonographic screening for major congenital anomalies is part of the standard obstetric care in many developed countries (5). In the United States, however, the American College of Obstetricians and Gynecologists recommends that sonographic evaluation of low-risk populations be reserved only for specific indications (6), such as an abnormal maternal serum screen (7). This recommendation is mostly made based on the uncertainty of whether an improvement in the survival of neonates with life-threatening anomalies can be expected from routine ultrasound of low risk pregnancies (8). It appears, however, that a large proportion of obstetricians in the United States order routine sonography for screening of congenital anomalies and the great majority of women in this country receive at least one sonographic evaluation during the course of their pregnancy.

Several controlled prospective studies have evaluated the possible impact of obstetric sonography on prenatal management, perinatal morbidity, and mortality. Studies from London and Trondheim failed to show any particular advantage with the use of routine sonography (9), while studies from Norway suggested that sonographic screening is associated with a reduction in the incidence of post term pregnancies, fetal death and low birth weight (10). In the Glasgow series, prenatal ultrasound screening allowed for more diagnoses of intrauterine growth retardation without an impact on clinical outcome (11). The Swedish multi-centric study showed a lower incidence of labor inductions due to post-term pregnancy and a lower incidence of low birth weight deliveries in patients who underwent routine sonography (12). This data shows that the controversy regarding the benefits of sonographic screening is far from resolved.

Another aspect of the controversy relates to its cost-effectiveness. Devore (13) estimated that the cost per anomaly detected by sonography was not greater than with routine use of maternal serum alpha-feto protein (MSAFP). Thus, if the routine use of MSAFP is justified, so is the use of routine sonography. To validate this observation, sonographic diagnosis of congenital anomalies would require a sensitivity of at least 60%. However, the sensitivity of sonographic screening is called into question by the poor results derived from the RADIUS study (14). Other studies have reported a much better diagnostic sensitivity (4).

Our data shows that the majority of detected renal anomalies were identified in patients without indications for a sonographic study other than routine screening in low-risk pregnancies. In 55% of these cases, the diagnosis was either unilateral or bilateral hydronephrosis, a condition that may benefit from modifications in prenatal management and can easily remain undetected at birth producing preventable morbidity. This supports the argument that detection of anomalies can best be achieved by routine sonography of low-risk patients. Taking into account that congenital anomalies represent 25% of all perinatal deaths and that 90% of them occur in patients without any risk factors, prenatal detection creates options for the parents.

After the prenatal diagnosis of certain congenital anomalies, the fetus may benefit from antenatal or early neonatal therapy and/or close surveillance of the newborn after being discharged home form the nursery. These potential benefits apply when renal anomalies are detected and these malformations are common (15). Because of their sonographic characteristics, a large number of them, especially those of an obstructive nature, are amenable for early sonographic identification. Many questions are raised when an antenatal diagnosis of hydronephrosis is made, including the possibility of *in utero* intervention, delivery route and the need for tertiary care (16). It also creates an indication for further testing with serial sonographic exams and invasive diagnostic studies such as amniocentesis. This not only may increase costs, but also significantly increase prenatal morbidity from excessive intervention, a factor that may be difficult to estimate.

Fetuses identified with renal collecting system dilatation *in utero* may benefit from post-natal evaluation. Persutte et al (15), reported that although post-natal surgery was necessary in only a small number of cases of prenatal hydronephrosis, surgical intervention was necessary in 33% of those patients found with vesico-ureteral reflux diagnosed prenatally. On the other hand, 25% of cases of isolated hydronephrosis resolve spontaneously during the first two years of life and thus, the prenatal diagnosis of these cases can lead to unnecessary intervention and parental anxiety (17). This may create several dilemmas when counseling patients. However, long term follow up is recommendable in the majority of cases of prenatally detected hydronephrosis, even after resolution, since recurrence is possible and renal damage may potentially be prevented (18).

It is still uncertain if prenatal diagnosis of congenital anomalies by ultrasound at its current stage can make a significant impact on perinatal outcome, although the results of this study warrant further evaluation by larger trials. Future studies must take into account not only perinatal mortality, but, also the potential reduction in long-term morbidity it may produce, compared to problems caused by unnecessary intervention. It is clear that relying on risk factors as indications for ultrasound studies will

not detect the majority of congenital anomalies, at least, those associated to the urogenital system, a group of malformations that may particularly benefit from prenatal detection.

Resumen

El propósito de este estudio es reportar la gama de anomalías congénitas renales identificadas sonográficamente y analizar las indicaciones para el estudio sonográfico que logró el diagnóstico. Todos los casos de anomalías renales diagnosticadas prenatalmente en nuestra institución de junio del 2001 a mayo del 2004 fueron evaluados retrospectivamente. Las indicaciones para la evaluación sonográfica fueron revisadas. Los casos se dividieron entre aquellos referidos para tamizaje sonográfico rutinario y aquellos referidos por otras indicaciones. Se identificaron 117 casos de anomalías renales congénitas y en 14 de los casos (11.9%), otras anomalías estaban presente. La hidronefrosis fue el diagnóstico más común, (64 casos) para una incidencia de 54.7%. Se identificaron 21 anomalías renales letales (17.9%).Riñones displásticos multiquísticos fueron identificados en 21 casos (17.9%). La agénesis renal se identificó en 15 casos (12.8%). Otras anomalías detectadas fueron: obstrucción a la salida de la vejiga 15 (12.8%), riñones ecogénicos 13 (11.1%), implantación ureteral ectópica 2 (1.7%), quistes renales 1 (0.8%), riñon pélvico 3 (2.6%), sistema colector doble 1 (0.8%) y riñon atrófico unilateral 1 (0.8%). En un total de 94 casos (80.3%) la única indicación pára evaluación sonográfica fue la de un tamizaje de rutina. El depender de factores de riesgo como indicación para estudios sonográficos no detectará la mayoría de las anomalías congénitas, especialmente aquella asociadas al sistema urogenital, un grupo de condiciones que particularmente se beneficiarian de detección prenatal.

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