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WAGR Complex

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Various epidemiological studies have shown aniridia to be increasingly associated with Wilms' tumor(1,2). The association of sporadic Aniridia with Genitourinary anomalies and mental retardation particularly makes a child high risk for the development of Wilms' tumor. Association of these four entities have been given the acronym WAGR complex. Only 29 patients of this complex were described till 1986(3). A few more have been added since then(4). We report a case of WAGR complex with Stage V Wilms' tumor treated with multimodal therapy.

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Case Report

A 2½-year-old boy was admitted to the Department of Pediatric Surgery, SPMCHI, Jaipur with complaints of asymptomatic mass abdomen for three months duration. There was no history of weight loss, hematuria, *etc.* Examination revealed anemia, bilateral hypoplasia of iris, obvious cranio-facial dysmorphism with abnormally large pinnae (*Fig. 1*). Nys- severe photophobia was noted. The child was severely mentally retarded. Abdominal examination revealed 10 × 8 cm firm retroperitoneal mass in the right lumbar region. The left kidney was also palpable. Examination of genitalia revealed severe penoscrotal hypospadias with right impalpable testis. A clinical diagnosis of WAGR complex was made with a likelihood of

Fig. 1. Photograph of the child. Note the large pinnae and obvious cranio-facial dysmorphism. The child has closed his eyes due to severe photophobia.

tagmus with bilateral Wilms' tumor involvement. There was no familial history of aniridia or Wilms' tumor. Routine investigations revealed a low hematocrit while blood urea serum creatinine and urinanalysis were essentially normal. Ultrasonography of the abdomen revealed bilateral renal masses. IVP revealed no renal function on the right side whereas there was distortion of pelvicalyceal system seen on the left side (*Fig. 2*). Buccal smears were negative for Barr's body and karyotyping revealed the chromosomal pattern to be 46 XY. Specific chromosomal study for 11p13 deletion could not be done due to non-availability of sophisticated techniques.

After initial build up, the child was taken up for exploratory laparatomy which revealed bilateral Wilms' tumor. On the right side, the kidney was wholly replaced by the tumor and was completely excised. The tumor was limited to kidney and there was no evidence of regional extension of tumor or any evidence of vascular involvement. There was lower polar involvement of the left kidney for which partial nephrectomy was done. No lymph node enlargement was noticed. Histopathological examination of tissues taken from both kidneys revealed bilateral Wilms' tumor with favorable histology.

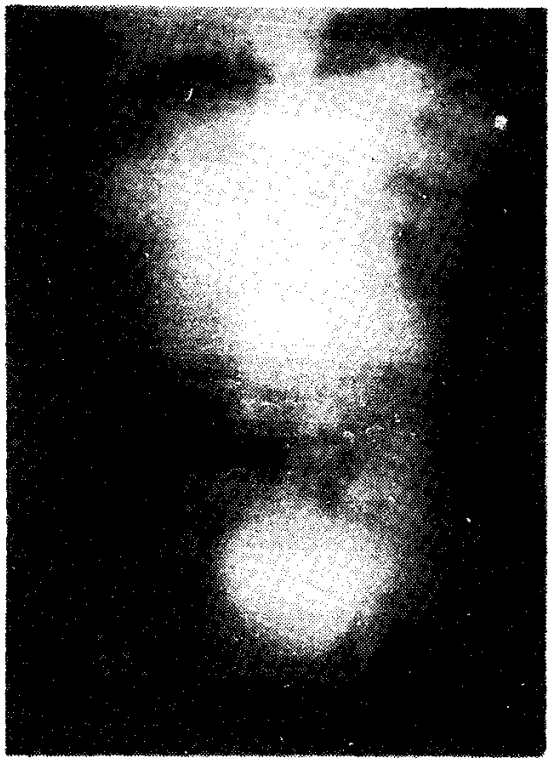

Fig. 2. IVP film showing non-functioning right kidney replaced by tumor and mild distortion of the lower calyx of the left kidney.

Child has a very stormy post-operative period. He required a re-operation after 7 days for post-adhesive intestinal obstruction. Chemotherapeutic drugs included actinomycin-D and vincristine, which were not well tolerated initially. On subsequent follow-ups, child could respond better to the chemotherapy regimen. No radiotherapy was given. After one year of initial surgery, clinical examination and investigations did not reveal any residual disease or distant metastasis. We plan to pex the right testis followed by repair of hypospadias.

Discussion

As many as 34 out of 100 patients of sporadic aniridia have been quoted to develop Wilms' tumor(5). The incidence is alarming if we compare this to the incidence of Wilms' tumor in general population which is about 7.8 per million(6). The aniridia-Wilms' tumor association was first noted by Miller and associates in 1964. They later enumerated the following features of a fully expressed aniridia – Wilms' tumor syndrome: development of tumor before 3 years, cogenital eye lesions, mental retardation, genitourinary anomalies, deformities of the pinna, and less commonly skull or cranio-facial dysmorphism, umbilical and inguinal hernias and hypo-

tonia(7). Riccardi *et al.* recognized the association of AGR triad (Aniridia, Genitourinary anomalies and mental retardation) to Wilms' tumor and thus helped in coining the term WAGR complexs(8).

Aniridia is in fact a misnomer, as complete absence of iris is never seen; instead there is bilateral hypoplasia of iris stroma(9). A similar finding was noted in our case also. Occasionally, this is associated with other ocular abnormalities such as cataracts, nystagmus, glaucoma, hypoplasia of macula including that of fovea centralis, micro-ophthalmia, ectopic lentis and refractory errors(9). None of these ocular lesions were seen in the present case.

WAGR complex gained importance as it was found to be associated with a deletion at 11p13(8). This stimulated considerable research regarding the role of genetic factor in the etiology of Wilms' tumor. Subsequent research, however, showed that more than one gene may be involved and 11p13 deletion may be a cytogenic observation specific to few patients such as those with WAGR complex(10). On the other hand, one patient with WAGR complex has been reported to have normal chromosomal pattern(11).

Through this report, we wish to highlight that the patients with WAGR triad have significant risk of developing Wilms' tumor and suggest frequent physical examinations and non invasive investigation like ultrasonography to pick up the tumor early. Early intervention even in cases of bilateral Wilms' tumor have a satisfactory outcome using multimodal therapy(12).

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