

## Case Report-1

# Wilms Tumour with Aniridia: A Case Report

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### ABSTRACT

A 19 month old girl was diagnosed to have Wilms tumour. She was also detected to have bilateral aniridia. There were no other congenital anomalies. Her twin sister also has bilateral aniridia, but no evidence of Wilms tumour. The child was treated with neoadjuvant chemotherapy followed by surgery. She continues to be alive and disease-free at 30 months of follow up.

### INTRODUCTION

Wilms tumor is the most common childhood renal tumor accounting for about 6% of all paediatric malignant diseases.<sup>1</sup> There is increased incidence of Wilms tumour amongst siblings and twins suggesting a possible genetic cause.<sup>2</sup> About 15% of children with Wilms tumour have associated congenital anomalies like aniridia (1.1%), hemihypertrophy (2.9%), musculo-skeletal (2.9%) and genito-urinary (4.4%) anomalies.<sup>3</sup> Aniridia is an uncommon congenital, bilateral panocular disorder in which various ocular structures like cornea, anterior chamber, lens, retina and optic nerve may be affected. Apart from ocular pathologies aniridia may be associated with certain systemic diseases like Wilms tumour, genito-urinary anomalies and mental retardation. We report a case of aniridia with Wilms tumour in one of twin sisters, where aniridia was present in both the sisters but Wilms tumour in only one.

### CASE

A 19-month-old girl presented with abdominal swelling of 4-month duration. She is one of twin

sisters delivered normally at term. The twins were considered monozygotic because of their phenotypic similarity. On examination she was found to have bilateral aniridia, non-tender right lumbar swelling with normal physical and mental development. Other than aniridia there was no congenital anomaly. There was no family history of aniridia or renal tumours. CT scan abdomen showed 70mm x 43mm x 36mm well defined heterogenous mass in superolateral aspect of right kidney, with the left kidney being normal. She had a normal chest skiagram. Fine needle aspiration cytology from the right renal mass was suggestive of Wilms tumour. Cytogenetics showed normal karyotyping (46 xx). Ophthalmological examination revealed bilateral aniridia with no other abnormalities (Fig below.) The other sister was also examined and was found to have bilateral aniridia but without any evidence (clinical and radiological) of Wilms tumour.



The child with Wilms tumour was treated with primary chemotherapy Vincristine, actinomycin-D and adriamycin (VCR+ActD+Adr) followed by right radical nephrectomy and adjuvant chemotherapy Ifosphamide + VP-16 (Etoposide) (IFS+ VP-16). Post-operative

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histopathological report was suggestive of Wilms tumour (Favourable histology). She completed treatment in May 2002 and is doing well for the last two and half years. Her twin sister is kept under close observation for early detection of Wilms tumour.

## DISCUSSION

The estimated relative risk of Wilms tumour in the co-twin is 250 times than in the normal population.<sup>4</sup> Juberg et al<sup>5</sup> reported the case of a pair of monozygotic twins in which the first child developed Wilms tumour at the age of 2 months while the other twin developed it at 1 year. They considered the twins to be monozygotic because of their phenotypic similarity. Maurer et al<sup>6</sup> reported 2 pairs of monozygotic twins with congenital anomalies and discordance for Wilms tumour. In one of the pairs of twins, aniridia with psychomotor retardation was present in both the children but Wilms tumour was present in only one child. In the other pair hemihypertrophy and Wilms tumour was present in only one child with the other child being completely normal. Although patients with sporadic aniridia are at risk of developing Wilms tumour, familial aniridia associated with Wilms tumour is also reported.<sup>7</sup> Risk of Wilms tumour is estimated to be 67 times higher if the child is already known to have sporadic aniridia.<sup>8</sup> Mutations in the transcription factor PAX 6 have been shown to be the cause of sporadic aniridia.<sup>9</sup>

The incidence of aniridia is approximately 1 in 50,000 in the general population.<sup>10</sup> One fifth of sporadic aniridia patients may develop Wilms tumour. Nanda et al reported 2 cases of Wilms tumour with sporadic aniridia, both presenting at a relative younger age.<sup>10</sup>

In our case both the sisters presented with aniridia but Wilms tumour was present in only one and that too in a relatively younger age. The

twins were identical and the chromosomal study did not reveal any abnormality thereby ruling out WAGR syndrome. (Wilms tumour, Aniridia, genitourinary malformations, and mental retardation) the importance of our case lies in the close observation of the other twin (with only aniridia) who is at risk of developing Wilms tumour. The high association of sporadic aniridia to Wilms tumour mandates a thorough ocular examination at birth along with renal ultrasonography, which is to be repeated every 3-6 months.<sup>7</sup> Conversely all the cases of Wilms tumour should be subjected to ocular examination for detection of any associated ocular anomalies.

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