678

Mascarenhas de Souza F[1], Carvalho Leão Barretto N J[1], Rebouças Prates R[1], Leão Barretto de Souza N C[1]
1. *Hospital Santo Antonio Obras Sociais Irmã Dulce*

OCHÔA SINDROME: A BRAZILIAN GIPSY COMMUNITY CASE SERIES AFTER 20 YEARS

Hypothesis / aims of study

The Ochôa Sindrome might have a severe urinary tract involvement and seems to be more prevalent in children in which parents there is some degree of consanguinity. This disease is a well described clinical setting in which the association of a neurogenic bladder with an abnormal facial expression found with the patients seeming to be laughing when they are crying and vice-versa. If the diagnosis and treatment are delayed the patients might have a poor outcome with a continuous upper urinary tract deterioration [1], [2], [3]. We describe a case series of Ochôa Sindrome with special attention to a Gipsy community in Brazil.

Study design, materials and methods

A retrospective chart review was made on 7 patients known to have Ochoa Sindrome in a Pediatric Urology section of a general hospital from january of 1988 to february of de 2008. Data on patient age, gender, age of diagnosis, image and urodynamic exams, first line treatment and subsequent treatments and outcome were seeked as well as their current status that was evaluated on a recent office visit with history and physical exam.

Results

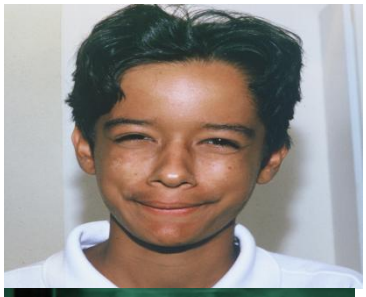
Case series: Seven patients, four male, were analyzed. Four of them belong to a gipsy community. Two of them are sibilings and two others are their cousins. Five patiens in this series know to have their patients with some degree of consanguinity. The first urology visit varied from one to 10 years but the diagnosis was not always promptly made since the face aspect was not recognized at first in five patients. Every patient had at least one febrile urinary tract infection and all but one had a weak and intermitent urinay stream with an important quality of life compromise. One patient was submitted to early vesicostomy. The first ultrasound showed bilateral hydronefrosis in five patients and the urethrocystogram showed bladder pseudodyverticulum in all and unilateral vesicoureteral reflux in five of them. Every patient received oxybutinin and 4 were put on clean intermittent catheterization. Two patients were submitted to bladder augmentation, three were told to have their bladder augmented, one is only 4 years old and has a vesicostomy and only one is continent and has a normal upper urinary tract.

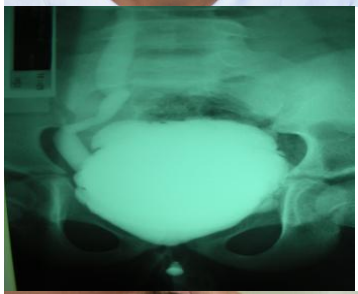
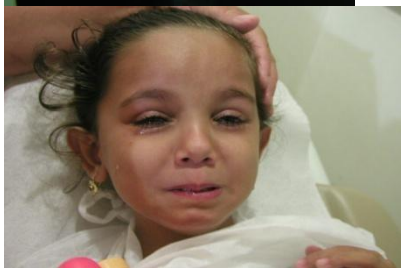
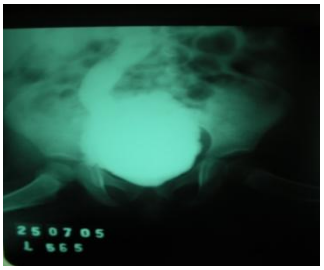
Interpretation of results

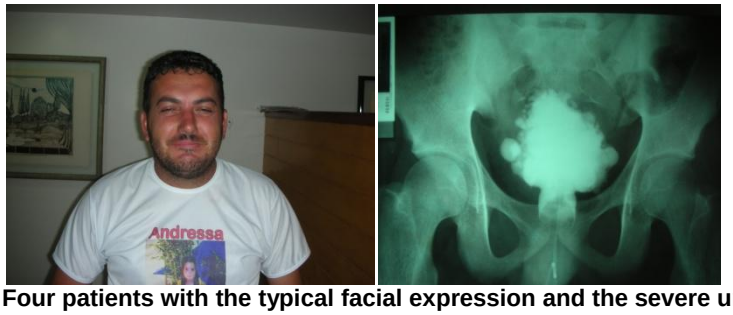
Ochôa Síndrome is a disease with a severe lower and secondary upper urinary tract involvement with early lower urinary tract symptoms and subsequent pyelonephritis, which often comes to a bladder augmentation need .

Concluding message

Early diagnosis and close follow up is the key point to a successfull outcome.





Four patients with the typical facial expression and the severe urinary tract changes present in Ochoa Syndrome.

References

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Is this a clinical trial?	**Yes**
Is this study registered in a public clinical trials registry?	**No**
What were the subjects in the study?	**HUMAN**
Was this study approved by an ethics committee?	**Yes**
Specify Name of Ethics Committee	**Comitê de Ensino e Pesquisa da Escola Bahiana de Medicina**
Was the Declaration of Helsinki followed?	**Yes**
Was informed consent obtained from the patients?	**Yes**