FREQUENCY OF INFANTILE PYLORIC STENOSIS AND OCULAR ABNORMALITIES IN TURNER’S SYNDROME IN BAHAWALPUR CITY

M. SIDDIQUE, SADIQ H. MALIK, TARIQ MAHMUD ARAIN, ASMA SHAUKAT, TABINDA YASMIN AND M. AHMAD SAJID

Departments of 1Pediatric Surgery, 2Pathology and 3Ophthalmology
Bahawal Victoria Hospital / Quaid-e-Azam Medical College, Bahawalpur
4Second Year, University of Liverpool. Liverpool. United Kingdom

ABSTRACT

Background: Turner’s syndrome is one of the most common of all chromosomal abnormalities. Pyloric stenosis is the most common pediatric surgical disorder of infancy that requires surgery for associated emesis. Ocular abnormalities are common in turner’s syndrome, but are underestimated and often neglected.

Materials and Methods: The study that was hospital – based observational, was conducted from September 1\textsuperscript{st}, 2005 to August 31, 2011 in the Al-Noor Hospital Yazman (Bahawalpur) in collaboration with ophthalmology department, Bahawal Victoria Hospital Bahawalpur. Laboratory investigations including electrolytes, blood gas analysis, and osmolality were done in pathology department, Quaid-e-Azam Medical College Bahawalpur.

Results: Frequency of pyloric stenosis in infants from 0 – 6 months 64.95% and infants 6 – 12 months is 35.09%, the ocular abnormalities amblyopia 19%, strabismus 23%, phoria only 43.5%, epicanthus 10%, hypertelorism 3%, ptosis 1%, nystagmus 0.5%.

Conclusions: There is a high frequency of ocular abnormalities and pyloric stenosis in infants with karyotype XO. Timely diagnosis may save from devastating effects of ocular complications leading to blindness and life threatening alkalosis due to pyloric stenosis in infants below one year of age.

INTRODUCTION

Turner’s syndrome is one of the most common of all chromosomal abnormalities, affecting 1 in 2000 live female births. The syndrome is best known for XO Karyotype, more accurately described as 45 X. Pyloric stenosis in infants is a narrowing of the outlet of the stomach in infancy, causing severe vomiting. Pyloric stenosis is the most common pediatric surgical disorder of infancy that requires surgery for associated emesis. Hypertrophic pyloric stenosis is commonly encountered in pediatric practice. The typical infant presents with non bilious projectile vomiting and dehydration (with hyperchloremic metabolic alkalosis) if the diagnosis is delayed it is life threatening. The infant usually presents at age of 3 – 6 weeks from birth. This condition accounts for one third of non bilious vomiting occurrence in infants and is the most common reason for laparotomy before age 1 year.

The frequency of hypertrophic pyloric stenosis is higher in turner’s syndrome. The first and the most important step in patient workup of suspected hypertrophic pyloric stenosis is through physical examination and ultrasonography recommended which has sensitivity and specificity close to 100% and laparotomy is done to correct hypertrophic pyloric stenosis. Ocular abnormalities are common in turner’s syndrome, but are underestimated and often neglected. Very few studies have been published in ophtalmic literature. High frequency of ophthalmic morbidity in turner’s syndrome population has been studied. Though ametropia is very common (around 40%), this is probably adequately dealt with by community optometrists. However higher rates of amblyopia (almost 30%) and strabismus (33%) is of greater concern.

There is a danger that delayed recognition may be sight threatening. The high frequency of these ocular complications would argue for early systematic screening of children with Turner’s syndrome and thus early detection and treatment of its sight threatening sequele.

MATERIALS AND METHODS

The study that was hospital – based observational in type, was conducted from September 1\textsuperscript{st}, 2005 to August 31, 2011 in the Al-Noor Hospital Yazman (Bahawalpur) in collaboration with ophthalmology department, Bahawal Victoria Hospital Bahawalpur. Laboratory investigations including electrolytes, blood gas analysis, and osmolality were done in...
pathology department, Quaid-e-Azam Medical College Bahawalpur.

Fifty seven infants with XO karyotype more accurately described as 45 X were included in study. Karyotyping of the patients were done from the Agha Khan University Hospital Karachi. Children above 1 year of age were excluded, similarly infants with pyloric stenosis without Karyotype XO were excluded from study. Ocular abnormalities in infants without 45 XO were excluded. All the study subjects were examined by ophthalmologists by fundoscopic and slit lamp examination.

All study subjects, after history taking were physically examined and ultrasonography was done to confirm the diagnosis of hypertrophic pyloric stenosis. Verbal consent was taken from the parents and the study was fully explained to them. All the expenses were borne by the researchers. The study was approved by the ethical committee of Quaid-e-Azam Medical College, Bahawalpur.

The blood gas analysis to confirm or otherwise, metabolic alkalosis was done by (Boehrigner) blood gas analyser. Electrolytes were performed by ISE (Medica) and osmolality calculated was done to confirm, dehydration.

**Statistical Analysis**

Percentage was calculated.

<table>
<thead>
<tr>
<th>(Age) Months</th>
<th>Number of patients Turner's syndrome (n = 51)</th>
<th>Frequency of Pyloric stenosis (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>0 – 6</td>
<td>37</td>
<td>64.91</td>
</tr>
<tr>
<td>6 – 12</td>
<td>20</td>
<td>35.09</td>
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**RESULTS**

In all 57 study subjects the sex chromatin in buccal smear was negative. The Karyotype in the peripheral blood was 45 – XO.

**DISCUSSION**

The birth rate of Bahawalpur City is increasing at a rate of 4.5 per year according to statistics. There were 1 Lac, 9 thousand 600 and 60 live births in the last year. The prevalence of pyloric stenosis in general population throughout world is 2 – 5 / 1000 live births.

In our study only infants with Karyotype XO were included so the frequency is much lower. In another study carried out on babies with Turner’s syndrome the ocular complications familial strabismus was most prominent ocular abnormality presents in 33% of the patients. Other eye findings in children ptosis (16%) hypertelorism (10%) epicanthus (10%) were found.

The frequency of ocular abnormalities were different in our study because we included only infants with karyotype 45 Xo, while in the study mentioned above karyotype 45 Xo, 45 Xo / 46 XX (mosaicism) were also included. That might be the cause of the difference between two studies. While in another study the frequency of ocular abnormalities was much greater than found in our study. The difference may be due to the fact that children above 1 year were also included in the study.

In a study carried on children it was found that the frequency of pyloric stenosis was much higher than ours. The difference is due to the fact that our subjects included in the study were only those who had karyotype 45 XO and with ocular abnormalities. The literature shows that there are only a few studies carried out on Turner’s syndrome infants who have pyloric stenosis and ocular abnormalities. The delay in diagnosis may lead to higher morbidity or mortality. Pyloric stenosis is a manifestation which can be diagnosed and treated relatively earlier as compared to ocular abnormalities which if goes unnoticed may lead to blindness.

It is concluded that frequency of pyloric stenosis in infants from 0 – 6 months 64.95% and infants 6-12 months is 35.09%, the ocular abnormalities amblyopia 19%, strabismus 23%, phoria only 43.5%, Epicanthus 10%, hypertelorism 3%, ptosis 1%, and nystagmus 0.5%.

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