



TURNERS SYNDROME IN A 38 WEEKS OLD FEMALE NEWBORN CHILD :A CASE REPORT.

Anatomy

Dr. Debajani Deka* Anatomy Department Assam *Corresponding Author

Dr. Kapil Kashyap Bora Anatomy Department Assam

KEYWORDS

INTRODUCTION:

Turners Syndrome is a condition of monosomy for X chromosome and total number of chromosome is 45 with karyotype 45, XO. It is one of the common type of aneuploidy among humans and is present in 1:2000 newborns with female phenotype. 45, XO karyotype is found in 50-60% of cases of turners syndrome and in rest of the case we may get 46XX or 46XY.

Phenotype of the Turners syndrome is characterized by short stature, webbed neck, flat chest, wide spaced nipple with sometimes ambiguous genitalia. Turners syndrome is often associated with congenital malformation such as streak ovary, and coarctation of aorta¹. In girls, older than 4 months and younger than 16 years of age, are found to have a significant decline in % of ovarian volume. If Y specific sequences are present then there may be virilization during puberty. Pregnancy is possible in 2% of cases if Xq13-Xq26 region is spared or if 46XX cell line is found⁴.

CASE REPORT: A 38 weeks old female newborn baby was found to be admitted in the department of pediatrics neonatal unit having birth weight 3.3 kg. There is history of refusal to breast feed, 2 episodes hypoglycemic seizure, umbilical discharge, respiratory distress with RR-48/min and HR-130/min. Regarding maternal history mother is 24 years old with Gravida 3 parity 3. There is a history of intrauterine death and a postnatal death in 3 months after delivery. The baby with turners syndrome has been found with features of widely spaced nipples, webbed neck, hypertelorism, bilateral lymphedema.

Ultrasonography-normal

Echocardiography-normal.

Karyotyping-45XO.

REVIEW OF LITERATURES:

According to Lilian Pinero Eca Alexis Boundedo Gauder (2009) a gene called SHOX is located in Xp22 and Yp11.3 in pseudoautosomal region of sex chromosome whose haploinsufficiency is believed to be responsible for short stature and other skeletal abnormality.

According to Ramesh K Sharma (2014) orbital hypertelorism resulted in orbits widely spaced apart resulting in appearance of broad nose. It gets established in 28mm embryo. Reason may be early ossification of lesser wing of sphenoid bones.

According to MMA Faridi, P Dhingra (2013) internipple index - internipple distance multiplied by 100 divided by chest circumference is the method of measuring nipple distance. Nipple can be considered widely spaced if >9.5cm apart and narrow spaced if <7.3cm.

DISCUSSION:

TABLE-1

AUTHORS	YEAR OF STUDY	FINDINGS
RAMESH K. SHARMA	2014	EARLY OSSIFICATION OF LESSER WING OF SPHENOID - HYPERTELORISM
PRESENT STUDY	2016	HYPERTELORISM

From the above discussion the newborn is found to have hypertelorism that is widely spaced eyes that is coinciding with clinical finding of Ramesh K. Sharma in 2014 in his case report.

TABLE-2

AUTHORS	YEAR OF STUDY	FINDINGS
P DHINGRA	2013	WIDELY SPACED NIPPLE, INTERNIPPLE DISTANCE > 9.5CM
PRESENT STUDY	2016	WIDELY SPACED NIPPLE

From the above discussion the child is found to have widely spaced nipple which is coinciding with clinical findings of PDhingra in his case report published in 2013.

TABLE-3

AUTHORS	YEAR OF STUDY	FINDINGS
GILES ATTON, KRISTIANA GORTON.	2015	LYMPHEDEMA OF HANDS AND FEET > 60% OF NEONATE WITH TURNERS SYNDROME
PRESENT STUDY	2016	LYMPHEDEMA OF HANDS AND FEET

From the above discussion we have found that lymphedema of hands and feet is found in greater than 60% of neonate with turners syndrome that is coinciding with present findings.



Hypertelorism

Webbed Neck

Widely Spaced Nipple

TABLE-4

AUTHORS	YEAR OF STUDY	FINDINGS
Ham P, Haber, Michael Ranke	1999	Younger than 16 years and older than 4 months decrease in ovarian volume.
Present study	2016	Normal ovary at birth

Ham P, Haber, Michel Ranke in 1999 study has mentioned that there is decrease in ovarian volume from 4 months to 16 years of age group in turners syndrome where in this baby both ovaries are of normal volume.

CONCLUSION-We can conclude early diagnosis of female infertility and prompt intervention to resolve it.

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