INTRODUCTION
Goldenher syndrome is a rare disorder with an incidence rate of 1 per 5800 live birth with male:female ratio 3:2. It is presumed to be an inherited condition causing morphological abnormalities of the part developed from the first and second branchial arch during blastogenesis. Goldenher first described the case in 1952 as a disease that present as a combination of several anomalies such as dermal epibulbar tumors, periauricular appendices and malformation of ears. Gorlin et.al named this syndrome as oculoauriculovertebral dysplasia due to presence of additional vertebral anomalies. Exact etiology of this disease is not known. Most of the cases are sporadic, though autosomal recessive, autosomal dominant and multifactorial inheritance has also been suggested. Chromosomal analysis shows no abnormalities. In this report we presented a case of Goldenher Syndrome in a 5 days old newborn who presented with all the classical features except ocular involvement.

Keywords: Goldenher syndrome, preauricular tag and sinus, mandibular hypoplasia, vertebral anomalies, facial asymmetry, rib anomaly, absent radius.

ABSTRACT
In 1952 Goldenher described a case with triad of pre auricular tags, mandibular hypoplasia and ocular (epibulbar) dermoid and described the case as Goldenger Syndrome. Exact etiology of this disease is not known. Here we present a case of Goldenher syndrome in a 5 days old newborn who presented with all the classical features except ocular involvement.

Gorlin et.al named this syndrome as oculoauriculo-vertebral dysplasia due to presence of additional vertebral anomalies. Exact etiology of this disease is not known. Most of the cases are sporadic, though autosomal recessive, autosomal dominant and multifactorial inheritance has also been suggested. Chromosomal analysis shows no abnormalities. In this report we presented a case of Goldenger Syndrome in a 5 days old newborn who presented with all the classical features except ocular involvement.

Keywords: Goldenher syndrome, preauricular tag and sinus, mandibular hypoplasia, vertebral anomalies, facial asymmetry, rib anomaly, absent radius.
The presence of anomalies of the ear and of limbs are necessary for the diagnosis of this syndrome. In this article we are presenting a case of 5 day old neonate diagnosed as a case of Goldenher syndrome.

CASE HISTORY
A single, preterm baby (gestational age 36 weeks, male, LSCS, Birth weight-2.5Kg), cried immediately after birth, admitted in our institution on day 5 of life with multiple congenital anomalies.

ANTENATAL HISTORY
The mother was 26 years old, primigravida, apparently in good health, without a prior history of any major illness. Mother had a history of intake of some oral abortifacient during first trimester as per advised by medical practitioner (no documents available). But as the abortion did not occur, mother continued the pregnancy. Otherwise the antenatal period was uneventful. She had regular antenatal checkups, taken iron & folic acid tablets regularly. Ultrasonography done on 23 weeks of gestation which revealed major congenital anomalies. She had no history of any fever, rash, lymphadenopathy, radiation exposure or any exposure to cat during the antenatal period. Mother was a non smoker & non alcoholic.

INTRANATAL HISTORY
It was a preterm institutional delivery by LSCS due to premature rupture of membrane & baby cried immediately after birth without any assistance.

IMMEDIATE POSTNATAL PERIOD
Uneventful. Baby put to mothers breast within 1 hour of birth, sucking normally from mothers breast. There was no history of any jaundice, convulsion or feeding difficulty. Baby admitted in our hospital on day 5 of life due to presence of multiple congenital anomalies.

HEAD TO FOOT EXAMINATION
Head Circumference-33cm, anterior fontanel-normal CC-29cm, Length-45cm,

Weight - 2.26kg on day 5 of life.
EAR - Right ear-preauricular tag, sinus & pit; Left ear-normal
ORAL CAVITY - tongue tie, bifid tongue, right sided deviation of angle of mouth, with bifurcation of lip at right angle of mouth.
FACE- micrognathia.
NECK- short neck.
EYE - no obvious anomaly seen.
HAND- right hand deformed with short forearm, hypoplastic right thumb, radial deviation of wrist joint, flexion of elbow joint.
CHEST- normal
ABDOMEN- normal male genitalia.
LOWER LIMB - no obvious visible deformity
SPINE & CRANIUM - no obvious deformity.

GENERAL AND SYSTEMIC EXAMINATION
Vitals - HR-110/min, RR-34/min. No pallor, cyanosis, jaundice, clubbing or oedema.
CVS - S1 & S2 audible, no murmur. RESP-bilateral vesicular breath sound, no adventitious sound audible, no intercostal or subcostal retraction.
GI - abdomen is soft, no organomegally, and intestinal peristaltic sound-present.
Modified Ballard Scoring-score -32; approximately gestational age-37 weeks.
INVESTIGATIONS
Complete blood count Hb-16.4gm%, TLC-11300 (N49,L35,M10,E2,B4), Platelet-3.4lakh,MCV-111.7,MCH-17.2,RDW-17.2,Reticulocyte count-1%.
Echocardiography-Small PDA with left to right shunt,ostium secondum type ASD.
Ultrasonography -no visceral or cerebral abnormality, hypoplasia of right parotid gland & irregularity in right half of mandible seen.
Chest X-Ray- rib abnormality.
Hypoplastic right half of mandible, absent of radius in right forearm. There is also deformity present in the cervical vertebra.

DISCUSSION
The above findings of the baby clinches us to the diagnosis of Goldenher syndrome.though the physical findings of the baby have some similarities with Teachercollins syndrome, it has now considered to be a different entity ,with features most commonly affecting bilaterally and having no aural or ocular abnormality.\(^8\)
The abnormalities are fond to be unilateral in 85% of cases and bilateral in about 10-33% cases.\(^9\) In asymmetric involvement with right side more affected than left side.\(^10\) our reported case has similar involvements. The reported frequency of cardiovascular abnormality ranging from 5-58%.Our case also had cardiac lesion in the form of ASD. There are some association found between maternal intake of some drugs like retinoic acid, thalidomide etc., with development of Goldenher syndrome.\(^11\) In this case also there was some history of maternal drug intake though no documents were available. In Goldenher’s syndrome ocular anomalies specially bilateral dermoid present in 60% of cases and vertebral and ear abnormalities are present in 40% cases.\(^12\)
The characteristic vertebral, ear, cardiac, facial, rib, mandibular anomalies all were present in our case. but surprisingly we didn’t found any ocular abnormalities, which is reported in maximum cases in literature.

CONCLUSION
Goldenher syndrome can also be presented without any ocular anomaly as seen in our case.

REFERENCES


