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Prenatal diagnosis of congenital anomalies prevalence, patterns and perinatal outcome an experience in tertiary care centre

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Abstract

Aim: To study the prevalence and patterns of prenatally diagnosed fetal congenital anomalies.

Materials and methods: This investigation was conducted as a prospective cohort study in the department of Obstetrics and Gynaecology., All India Institute of medical Sciences (AIIMS) Raipur (CG), All antenatal women attending ANC at AIIMS, Raipur with their scan reports who were willing for regular follow up and giving consent to participate in the study were enrolled for the study over 1.5 years duration. All investigations were noted including double marker at 11 to 13+6 weeks gestation age (GA), ultrasonography anomaly scans at 11 to 13+6 weeks GA and TIFFA at 18-20 weeks GA and followed up till delivery.

Results A total of 3830 antenatal cases were enrolled amongst them 215 fetus were diagnosed with congenital anomalies. Amongst them two were twin pregnancies. Total birth during this period was 1429 including live birth and still births. Thus, the prevalence of congenital anomalies is 1518 per 10,000 live birth Accounting for 15.18% and 95.35% patient were diagnosed antenatally and 4.6% postnatally. Maximum congenital anomalies were diagnosed at 14-20 weeks of gestation i.e. 46.5%.

Introduction

Congenital anomalies, commonly referred to as birth defects, are structural and functional abnormalities in any organ system of the body. These abnormalities can be identified during pregnancy, delivery, or at a later stage in life. Approximately 240,000 infants perish globally within 28 days after being born each year as a result of congenital diseases. Congenital

abnormalities result in an additional 170,000 deaths of children aged 1 month to 5 years. The incidence of congenital anomalies is similar worldwide, with rates of approximately 3% in the United States3, 2.5% in India4, and 2% to 3% in the United Kingdom5. The most common conditions are congenital heart problems, orofacial cleft, Down syndrome, and neural tube defect. Congenital abnormalities account for 15% of prenatal mortality and 10% to 15% of newborn fatalities in India.

The use of preconceptional screening and diagnostic tests, such as non-invasive methods like ultrasonography, fetal 2D-echocardiogram, MRI, and maternal blood biochemical indicators, has significantly enhanced the ability to detect congenital defects before birth. Approximately 53% of Congenital abnormalities are currently being identified as early as 14 weeks gestational age (GA)9. Between 18 and 20 weeks of gestational age, targeted imaging for fetal anomaly (TIFFA) can detect between 60% to 90% of anomalies, depending on their nature. The inclusion of fetal echocardiography at 22 weeks of gestation in prenatal ultrasound screening has significantly improved the accuracy and effectiveness of diagnosing cardiac abnormalities in the womb.

Prenatal diagnosis of congenital anomalies facilitates informed decision-making for parents and clinicians regarding the management of the pregnancy. This includes the option of terminating the pregnancy or continuing with it, as well as effective planning for potential complications during labour, after birth, and the immediate management of postnatal birth defects. Additionally, it aids in identifying potential risk factors for future pregnancies. Therefore, accurately predicting and diagnosing congenital defects during the period around conception can serve as a valuable tool in decreasing both the morbidity and mortality rates during the perinatal period.

The data on the prevalence and patterns of congenital anomalies in India is scarce and no such studies are available from the state of Chhattisgarh. The present study is carried out in a tertiary care centre, which is the main referral centre for the Chhattisgarh state. Collecting data for predicting of problem on the basis of the medical, social and obstetric history and physical examination further detection of congenital anomalies by prenatal anomaly scans, biochemical marker and neonatal screening thus will be helpful in generating the data for the state of Chhattisgarh.

Objective

To study the prevalence and patterns of prenatally diagnosed fetal congenital anomalies.

Study subject: study involves human subjects only

Type of study: A prospective cohort study

Study setting: Department of Obstetrics and Gynecology, All India Institute of medical Sciences (AIIMS) Raipur (CG)

Study population:

Pregnant women attending antenatal clinic (ANC) at AIIMS Raipur

Inclusion criteria:

1. All antenatal women attending ANC at AIIMS, Raipur in early pregnancy (Before 11 weeks) of gestation
2. Women attending ANC in late pregnancies with their early/ late anomaly scan reports.
3. Willing for regular follow up in AIIMS, Raipur
4. Giving consent to participate in the study

Sample size:

Calculated by formula-

$$n_0 = \frac{Z^2 pq}{e^2}$$

Z=1.96(at 95% confidence interval)

P=2.5 (prevalence of congenital anomalies)[4] q=100 - p

e=absolute error= 2

sample size =$\frac{1.96 \times 1.96 \times 2.5 \times 97.5}{2 \times 2}$

=233

Considering lost to follow up of 10% the calculated sample size is 256

Duration of study-1.5 Years.

Methodology

All antenatal women attending ANC at AIIMS, Raipur in early pregnancy or women attending ANC in late pregnancies with their early/ late anomaly scan reports who were willing for regular follow up in AIIMS, Raipur and giving consent to participate in the study were enrolled for the study.

After taking a written informed consent a detailed history including personal profile, menstrual history, Obstetric history, past history, family history, history of present pregnancy, any exposure to teratogens, radiation or any chemical or substance abuse, infections during pregnancy etc. were taken.

A detailed general, systemic and obstetrical examination were performed.

All previous investigations including ultrasound, biochemical markers were noted and were subjected to further investigations and follow up as per the routine antenatal protocol of the institute, which includes routine blood investigations, double marker at 11 to 13+6 weeks gestation age (GA), ultrasonography anomaly scans at 11 to 13+6 weeks GA and TIFFA at 18-20 weeks GA. Fetal echocardiography was advised only when indicated (e.g. history of congenital heart disease, risk of trisomy 21 etc.). Regular follow up of enrolled antenatal women was done till delivery.

Detailed examination of the neonate/abortus was performed by the neonatologist with special reference to congenital anomalies which also included the required investigations for postnatal diagnosis & confirmation of anomalies.

The neonates were followed up for one month following delivery to assess the perinatal & early neonatal outcome. To ensure comprehensive documentation of all irregularities, the medical professionals in the obstetric/gynecology department were notified and furnished with pertinent details regarding the study.

The perinatal outcome were noted in form of spontaneous miscarriage, medical termination of pregnancy (MTP), preterm delivery, term delivery, live births, stillbirth, postnatal diagnosis of congenital anomalies, early neonatal mortality etc.

Statistical analysis

The data was inputted into the Excel spreadsheet. The prevalence of congenital anomalies was determined according to the guidelines provided by the World Health Organization birth defects surveillance manual 201411 and the European Surveillance of Congenital Anomalies (EUROCAT)12.

The overall incidence rate of congenital anomalies (per 10,000 births) was determined by dividing the total number of cases of congenital anomalies, which includes live births, stillbirths, and elective terminations of pregnancy for fetal anomaly (ETOPFA), by the total number of births, including both live and stillbirths.

The live birth prevalence (per 10,000 live births) was determined by dividing the number of live births with congenital abnormalities by the total number of live births.
A fetus or newborn with numerous malformations was included in each category of anomaly only once.

The prevalence of congenital anomalies diagnosed prenatally (per 1000 births) was determined by dividing the number of cases diagnosed before birth by the total number of births, including both live births and stillbirths.

The classification of major and minor congenital malformations was determined according to the guidelines outlined in the WHO birth defects surveillance manual[11]

Perinatal outcome was calculated as the proportion of perinatal deaths in babies with congenital anomalies and perinatal deaths in babies without congenital anomalies per thousand total births (live and still birth)

RESULTS

A total of 3830 new antenatal visited ANC at AIIMS, Raipur amongst them 215 fetus were diagnosed with some or other congenital anomalies. Amongst them two were twin pregnancies. Total birth during this period was 1429 including live birth and still births. Thus, in the present study the prevalence of congenital anomalies is 1518 per 10,000 live birth accounting for

15.18%. The mean age of the women with congenital anomaly fetus was 26.7 years. The eldest woman was 38 years (**Table 1**.). The prevalence of antenatally diagnosed congenital anomaly is143.45 per thousand birth and the prevalence of postnatally diagnosed congenital anomaly is 6.9 per thousand birth (**Table 2**.). In the present study maximum congenital anomalies were diagnosed at 14-20 weeks of gestation i.e. 46.5%(n=100). In 2.9% (n=6) anomalies detected at 1st trimester anomaly scan. However, a big number i.e.45.9% were diagnosed late in pregnancy i.e. after 20 weeks. In this study 4.9% (n=10) were diagnosed after birth (**Figure1**). Congenital anomalies was divided in two groups major and minor congenital anomalies, depending on the severity[12] (**Table.3**). Out of total 215, 95.8% (n=205) were isolated anomalies (only one system involved) and 4.19% (n=10) were with multiple (two or more systems involved) in fetus (**Table.4**). Multiple anomalies include Non-immune fetal hydrops 1.3% (n=3) followed by Ventriculomegaly with club foot 0.4% (n=1), Pierre Robinson syndrome (low set Ear, cleft v shaped palate, retrognathia, macroglossia, feet syndactyly of right toe,2, 3-digit, bilateral CTEV) 0.4% (n=1). Turner syndrome (Genu recruvetum, scoliosis, protuberant eye and short neck) 0.4% (n=1), situs inversus 0.4% (10%), Arthrogryposis Multiplex 0.4% (n=1%), Umbilical cord cyst +mega bladder+ bilateral renal pelvic dilatation 0.4% (n=1) and Syndromic baby (depressed nasal bridge, low set ear, high arched palate, bilateral inguinal hernia, hydrocele, micropenis, CTEV) 0.4% (n=1). Each anomaly was counted separately anomalies in each system for fetus having multiple anomalies accounting it to be n=223, as 10 fetus had multiple anomalies. Most common congenital found was genitourinary system 32.7% (n=73)followed by Cardiovascular/thoracic system 26.4% (n=59),central nervous system (CNS)14.7%(n=33), umbilical cord 7.1%(n=16), Musculoskeletal system 8.07% (n=16), head and neck 5.3% (n=12) ,Gastrointestinal system 2.13% (N=5), biochemical 2.13%(N=5), and least common was spinal cord 0.85% (n=1.86%(n=4) and facial (absent nasal bone) 2.23% (n=12) (**Figure 2**).

Table 1. Distribution of sociodemographic and obstetrics characteristics of women with congenital anomalies in fetus(N=215)

Sr no.	Age group	N	%
1	<18	0	0
2	18-25	89	41.4
3	25-40	126	58.6
4	>40	0	0
Sr No	Education	N	%
1	Illiterate	2	0.9
2	Primary School	0	0
3	High School	31	14.5
4	Graduate	175	81.4

5	Postgraduate	7	3.3
Sr No	Residential Status	N	%
1	Urban	210	97.7
2	Rural	5	2.3
Sr no	Gravida	N	%
1	1	122	56.7
2	2	71	33.02
3	3	19	8.83
4	4	2	0.93
5	5	1	0.46
	Total	215	100

Table 2. Distribution of congenital anomalies according to the time of diagnosis- antenatal or postnatal

Sr No	Time of Diagnosis	N (215)	%
1	Antenatal	205	95.35
2	After Birth	10	4.65
	Total	215	100

Table 3. Distribution according to major anomalies and minor anomalies

Sr No	Type of Congenital Anomaly	N (215)	%
1	Major	63	29.3
2	Minor	152	70.69
	Total	215	100

Table 4. Distribution of congenital anomalies according to isolated, multiple anomalies

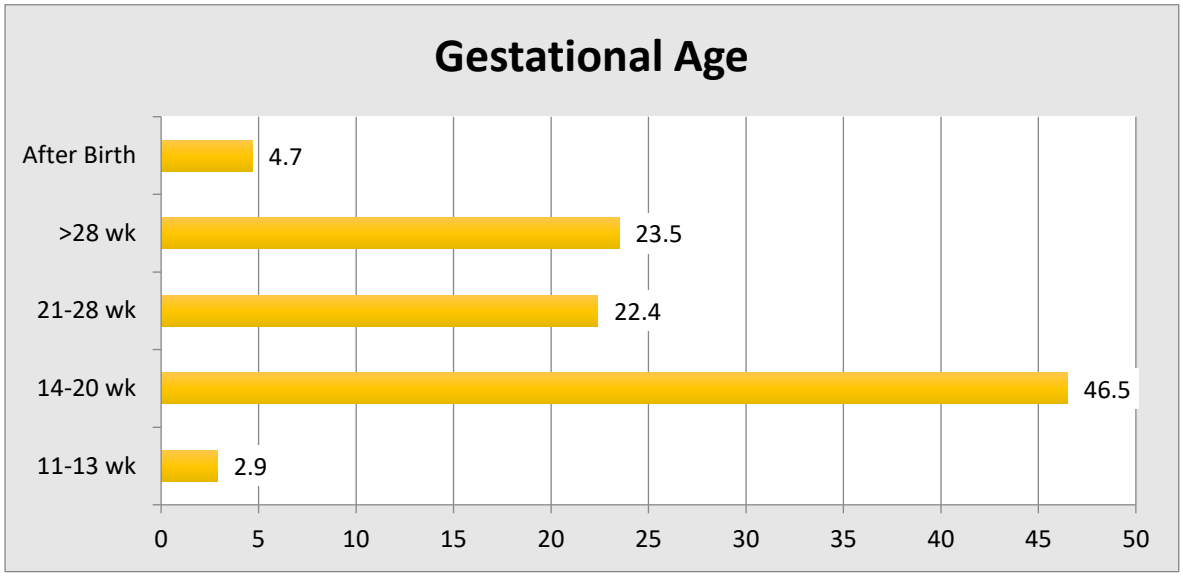

Fig 1. Demonstration according to Congenital Anomalies and Gestational Age at The Time of Diagnosis

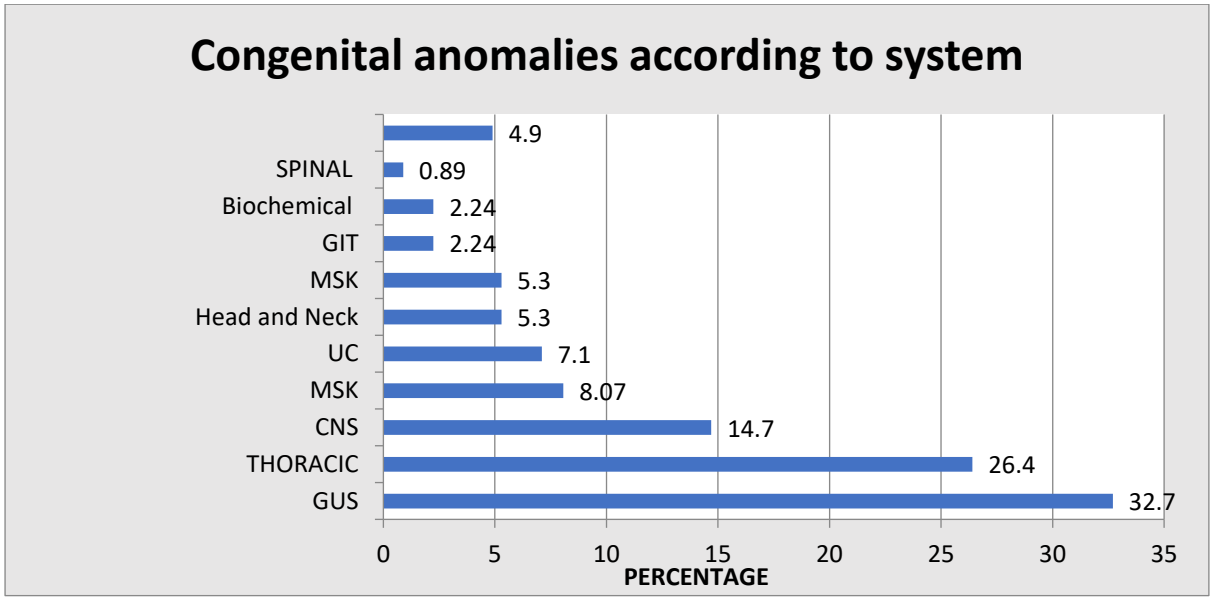

Fig 2. Demonstration of Congenital Anomalies According to System Involved

Sr No	Congenital Anomaly	N (215)	%
1	Isolated	205	95.34
2	Multiple	10	4.66
	Total	215	100

DISCUSSION

A total of 3830 new antenatal visited ANC at AIIMS, Raipur amongst them 215 fetus were diagnosed with some or other congenital anomalies amongst them two were twin's pregnancy. A total birth during this period was 1429 including live birth and still births.

The prevalence of congenital anomalies was 15.6% which is almost double than the reported by Akimoladun et al[13] (6.2%). Similarly, the prevalence of major congenital anomalies in our study is 4.4% which is higher than as reported by others Bhide et al[14] (2.3%), sallout et al[15] (2.7%), Rani et al[16] (0.9%) and Butt et al[17] (2.15%). The reason for the higher prevalence can be explained as our center is a referral tertiary care center and almost all the women from state are referred to AIIMS, due to availability of good antenatal, intrapartum and NICU facility. In this study the eldest woman was 38 years and youngest was 19 years. The study population was distributed in 4 age groups. The majority of the women 58.6% (n=126) were between 25-40 years followed by 41.45(n=89) in age group18-25 years, which indicates higher prevalence of congenital anomalies among women between age group 25-40 years and results are comparable to sallout et al[15] (26-35), Rani et al[16] (20-30) and Akinmoladun et al[13] (26 and 34 years).The reason for this may be because none of the women were in <18 years or > 40 years of age so, we don't have the data for the same. Majority of the women were primigravida 56.7%(n=122) followed by second gravida 33.02% (n=71). Rani et al[16] also found in her study that majority were nulliparous n= 25, sallout et al[15] found median parity to be 2. Out of total 215 women with congenital anomalous fetus 95.35% (n=205) patient were diagnosed antenatally and 4.9%(n=10) in postnatal period. Accounting to the prevalence of antenatally diagnosed as 143.45 per thousand birth which was compare to Rani et al[16] 94% antenatally and 6% after birth . Maximum congenital anomalies were diagnosed at 14-20 weeks of gestation i.e. 46.5%(n=100). In 2.9% (n=6) anomalies detected at 1st trimester anomaly scan. However, a big number i.e.45.9% were diagnosed late in pregnancy i.e. after 20 weeks. In this study 4.9% (n=10) were diagnosed after birth which was comparable to Rani et al[16], they divided gestation age at diagnosis as follows <28 weeks n=15(30%),28-37 weeks n=26 (52%),>37 weeks n=6(12%) and after birth n=3(6%). Sallout et al[15] found median gestational age to be 38 weeks with diagnosis of congenital anomaly in his study and again he described each system separately. Cranial 38 weeks, Neural tube defect 31week, Face 25 weeks, Thoracic 32 weeks, Cardiac 31weeks, Ventral wall defects 29 week, Abdominal 30.5-week, Genitourinary system 31 weeks, and Skeletal 35 weeks. Akinmoladun et al[13] divided gestation age of diagnosis in the following category and there results as follows detection in weeks 18-22 n= 22 (59.5), 22-28 n=9 (24.3) >28 n= 6 (16.2) .Drukker et al[18] found n= 103 (21.7%) were detected before the anomaly scan, n=174 (36.7%) at the anomaly scan,n= 11 (2.3%) after the anomaly scan and before the third-trimester scan=43 (9.1%) at the third-trimester scan and 143 (30.2%) after birth.In this study congenital anomalies was divided in two groups major and minor congenital anomalies, depending on the severity. Majority were minor congenital anomalies 74.9%(n=161) and 25.1%(n=161) were major congenital anomalies. In contrary Akinmoladun et al[13] found more major anomalies (59.6%) than minor anomalies, Fida et al[19] found major anomalies in 95.9% of their patients. Major Congenital anomalies show considerable variation all over the world with prevalence ranging from <1% to 8 %. The variations in the prevalence may result from differences in the study designs or may be the source of data, the length of

observation, and the methodology for definition and categorization of the malformations. 95.8% (n=205) were isolated anomalies (only one system involved) and 4.19% (n=10) were with multiple (two or more systems involved) in fetus. Multiple anomalies include Ventriculomegaly with club foot n=1(0.4%), Pierre Robinson syndrome (low set Ear, cleft v shaped palate, retrognathia, macroglossia, feet syndactyly of right toe,2, 3-digit, bilateral CTEV) n=1(0.4%), Non-immune fetal hydrops n=3(1.3%). Turner syndrome n=1(0.4%), situs inversus n=1(0.4%), Arthrogryposis Multiplex n=1(0.4%). Comparable study was done by sallout et al[15] in his study found out of 217 patients, n=71 (32.7%) had fetuses with complex anomalies and n=146 (67.3%) had fetuses with isolated anomalies. Each anomaly was counted separately anomalies in each system for fetus having multiple anomalies accounting it to be n=223, as 10 fetus had multiple anomalies. Most common congenital found was genitourinary system 32.7% (n=73) **(Table. 5)** and in genitourinary system renal pelvic dilatation was most common **(Table 6)**.

Table 5. Congenital anomalies as per the system involved reported by different authors

System involved	Present study	Sallout et al[15]	Rani et al[16]	Akinmoladin et al[13]	Gagnon et al[20]	Almeida et al[21]	Drukker et al[18]	Silesh et al[22]	Bhide et al[14]
CNS	14.7%	(28.6 %)	40%	24.3%	-	28.6%	10.1%	28.1%	11.9 %
Face and neck	5.3%	11.5%,	-	2.7%,	12.8%	-	4%	-	-
CVS/ Thoracic	26.4%	25.8%,	16%	13.5%	41.97%	12.5%	18.5%	14%	33.3%
GIT	2.13%	22.6%,	14%	16.2%	10.2%	14.11%	1.8%	20.6%	9.5%
Spine	0.85%	10.6%		-	8.3%	-	-		-
GUS	32.7%	38.6%,	8%	16.2%	19.3%	22.1%	20.4%	1.5%	23.8%
Muscu-loskele-tal system	8.07%	23.5%	40%	5.4%,	4.27%	6.45%	10.5%	16%	21.4%

Multis ystem/ Chrom osoma l	4.48	32.7	-	21.6%,	-	3.22	4.42%	15%	-

System involved	Most common anomaly in the system involved in Present study	Name of the authours and the most common anomaly found in their study
Central nervous system	Ventricular dilatation (5.8%)	Sallout et al[15] – hydrocephalus and ventriculomegaly Rani et al[16] - hydrocephalus Akimoudin et al [13] – Anencephaly
Head and neck	Cystic hygroma (1.7%)	Silesh et al [22] - clept lip Drukker et al[18] – clept lip
Cardiovascular/thoracic	Intracardiac echogenic foci (19.2%)	Rani et al[16], Silesh et al[22], Akimoudin et al [13]- Ventricular septal defect
Gastroinstestinal system	Duodenal atresia (0.89%)	Rani et al[16] & Silesh et al[22]- omphalocele Akimoudin et al[13]- exomplaos
Spine	Spina bifida (0.4%)	Rani et al[16]& Silesh et al [22]– spina bifida
Genitourinary system	Renal pelvic dilatation (27.8%)	Sallout et al[15] & Rani et al[16]- Hydronephrosis Akimoudin et al[13]- multi dysplastic kideny Silesh et al[22]- hypospadiasis
Musculoskeletal system	CTEV (4.9%)	Rani et al[16] – limb defect Silesh et al[22]- CTEV Akimoudin et al[13]-Thanatrophoric dyplasia and osteogenesis imperfecta Drukker et al[18]- skeltal dysplasia

Syndromes	Turner syndrome- 01 Pierre robin syndrome - 01 Unidentified syndrome - 01 Arthogryposis gryposis - 01	Silesh et al[22]- down syndrome 06 Cherian et al[23] - down syndrome 04, edward syndrome- 03, pierre robin syndrome 02, TAR syndrome 01, VACTREL 01, dysmorphic features 43.

Table 6–Most common congenital anomalies identified in different systems

Limitation

Our study is hospital based in the referral centre for the state of Chhattisgarh and also from nearby states (Orissa and Madhya Pradesh) so the study does not represent actual status of prevalence of congenital anomalies in Chhattisgarh alone

Conclusion

The commonest system involved was genitourinary followed by cardiovascular system. The majority of congenital anomalies were of unknown cause. Prenatal ultrasound aided in the timely identification and termination of pregnancies affected by congenital anomalies, hence decreasing the financial load and emotional distress experienced by the couple. Obstetrician, paediatric surgeon, paediatricians, geneticist, and radiologist were necessary to handle viable congenital abnormalities.

Conflict of interest: Author declares no conflict of interest

Ethical clearance: Consent was obtained or waived by all participants in this study. Institute Ethics Committee of All India Institute of Medical Sciences, Raipur issued approval AIIMSRPR/IEC/2019/355. At the convened meeting of IEC-AIIMS, Raipur held on 04.09.2018, the IEC voted to approve the above referenced protocol.

Animal Ethics: Animal subjects**:** All authors have confirmed that this study did not involve animal subjects or tissue.

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