

# Congenital Right Diaphragmatic Defects: Our Institutional Experience

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## Abstract

**Background:** The descriptive clinical study was conducted to analyse the clinical profile as well as the outcome of congenital right diaphragmatic defects among children including neonates in a tertiary care referral neonatal and paediatric centre in southern Karnataka, India. **Materials and Methods:** This retrospective and prospective observational clinical study was conducted from January 2005 to August 2019, over a period of 14.7 years in a tertiary care referral neonatal and paediatric centre. Clinical characteristics and risk factors of 33 children including neonates admitted and diagnosed with congenital right diaphragmatic defects were assessed both pre- and postoperatively. Neonates and children with acquired right diaphragmatic hernia defects and the left-sided diaphragmatic defects were not included in this clinical study. **Results:** For statistical as well as clinical analysis, 33 study subjects were grouped into four groups, depending on the pre-operative and intraoperative findings as well as on their final diagnosis. Group I comprised right congenital diaphragmatic hernia (RCDH) ( $n = 18$ ), Group II comprised RCDH with sac ( $n = 6$ ), the babies with diagnosis of right diaphragmatic eventration were included in Group III ( $n = 7$ ), whereas babies with other right-sided diaphragmatic hernia defects diagnosis were included in Group IV ( $n = 2$ ). **Conclusion:** Right-sided congenital diaphragmatic defects, though rare, do carry excellent survival if referred early and managed in a tertiary care neonatal and paediatric centre as that of left diaphragmatic defects.

**Keywords:** Diaphragmatic, hernia, neonates, right sided and congenital

## INTRODUCTION

Congenital diaphragmatic hernia (CDH) occurs in 1 in 2000–4000 live births and accounts for 8% of all major congenital anomalies. CDH is a major surgical emergency in newborns because the key to survival depends on the prompt diagnosis and treatment. Hence, the CDH may be of three basic types and those are posterolateral Bochdalek hernia, the anterior Morgagni hernia and the hiatal hernia. Most common type left-sided Bochdalek hernia contains large and the small bowel with or without intra-abdominal solid organ, whereas right-sided hernia (incidence 13%) contains only the liver with or without bowel tend to be herniated into the thorax.

## MATERIALS AND METHODS

This was a combination of retrospective and prospective observational clinical cohort study conducted from January 2005 to August 2019 over a period of 14.7 years at a tertiary

care neonatal referral centre in Karnataka, India. The data were collected from Neonatal Intensive Care Unit (NICU) medical records, paediatric surgical registry and the questionnaire put forth to the parents or caregivers. Children including neonates from day 0 to 30 referred with a clinical diagnosis of right congenital diaphragmatic defects were included in this study. The diagnoses of right sided diaphragmatic defects were made from chest roentgenography and ultrasonography [Figure 1a]. Contrast-enhanced computerised tomography and upper gastrointestinal dye studies were done in clinically doubtful cases. Depending on the pre-operative and intraoperative diagnosis, they were grouped into four groups. Group I included right-sided diaphragmatic hernia. Group II included right diaphragmatic hernia with the presence of sac. Group III included right diaphragmatic eventration and Group IV included

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other right sided defects like ,retro sternal /para esophageal hernia and Morgagni hernia.

Exclusion criteria included neonates and children with left diaphragmatic defects and acquired right diaphragmatic defects were excluded from this clinical study. Neonates and children with left diaphragmatic defects and acquired right diaphragmatic defects were excluded from this clinical study.

Repair of right-sided diaphragmatic defects under general anaesthesia was adopted as treatment modality after initial medical stabilisation. Assisted ventilation was provided for neonates requiring ventilation both pre- and postoperatively. Surgery was performed when the neonate’s general condition improved and blood gases analyses were stabilised for at least 24 h. Cardiac malformations were diagnosed using two-dimensional (2D) echocardiography. Persistent pulmonary hypertension of the newborn (PPHN) was defied as pre- and post-ductal oxygen saturation difference >10% and was confirmed by 2D echocardiography.

Medical stabilisation was defined by the following criteria (a) normal haemodynamic variables: mean blood pressure >40 mmHg, (b) disappearance of pre- or post-ductal saturation difference and the signs of PPHN on 2D echo, (c) switch to controlled mode of ventilation well tolerated with moderate values of peak inspiratory pressure (15–20 cm H<sub>2</sub>O) and adequate oxygenation achieved with FiO<sub>2</sub> ≤0.4. Oxygenation index (OI) was calculated by the following formula: (mean airway pressure [MAP] × FiO<sub>2</sub> × 100)/post-ductal O<sub>2</sub>, where MAP is MAP. FiO<sub>2</sub> is fractional inspired oxygen and PaO<sub>2</sub> partial pressure of oxygen in arterial blood.

The Institutional Ethics Committee has approved this study.

### Statistical analysis

The following factors were statistically analysed such as gestational age in weeks, birth weight in grams, age on admission, gender, the presence of cardiac malformations,

PPHN, requirement of pre- and post-operative ventilation, weight at the time of surgery, modes of surgery and duration of hospital stay among grouped subjects. Continuous variables were reported using mean ± standard deviation, median and interquartile ranges. Categorical variables were reported using frequencies and percentages. Student’s *t*-test and Chi-square test were used to find the association between variables. ANOVA or Kruskal–Wallis test were used to assess statistical significance based on the distribution of variables among groups [Table 1].

All the statistical analyses were performed using SPSS software version 18.0 (IBM, New York: Routledge, USA).

## RESULTS

A total of 33 children including neonates with right diaphragmatic defects were enrolled in this study. 32/33 (96.96%) underwent various surgical procedures. 1/33 (3%) baby expired preoperatively. Of those who underwent surgery, 31/33 (93.93%) survived and 2/33 (6.25%) neonates expired postoperatively. One baby expired preoperatively (1/33, [3.03%]).

The study subjects were grouped for statistically evaluation into four groups depending on pre-operative evaluation and intraoperative findings and histological findings. Group I included right diaphragmatic hernia, Group II included right diaphragmatic hernia with sac, Group III included right diaphragmatic eventration and Group IV included other defects of right diaphragm.

The mean age at presentation (days) to the hospital for surgery: Group I had a mean of 244 ± 353.8 (median: 4, range: 1–1080), Group II had a mean of 403 ± 624.5 (median: 165, range: 2–1640), Group III had a mean of 270 ± 374.8 (median: 180, range: 7–1095) and Group IV had two patients with a mean of 112.5 ± 95.46 days.

In Group I, male were 11 (33.33%) and 7 (21.21%) were female with a ratio of 3:1, Group II had 3 (9.09%) males and 3 (9.09%) females with a ratio of 1:1, Group III included 4 (12.12%) males and 3 (9.09%) females with a ratio of 3:2 and Group IV had one male and female each (3.03%).

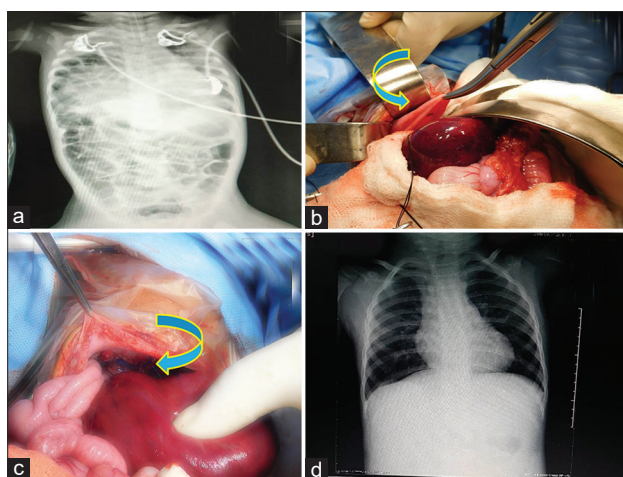


Figure 1: Clinical images of right CDH. (a) Pre-operative chest X-ray. (b) Intraoperative appearance of right CDH with sac. Blue arrow pointing at sac. (c) Blue arrow pointing at repaired diaphragmatic defect. (d) Post-operative chest X-ray. CDH: Congenital diaphragmatic hernia

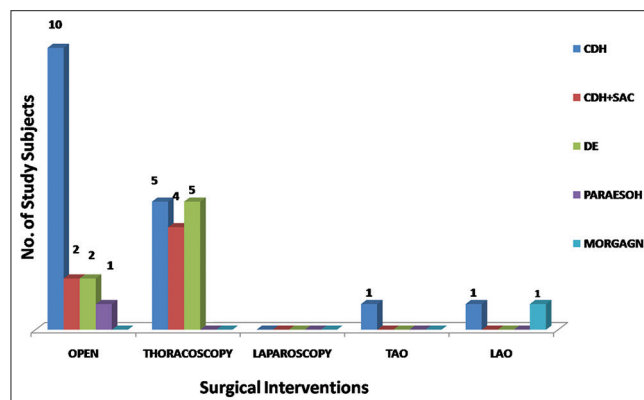


Figure 2: Graphical representation of modes of surgical interventions

**Table 1: Comparison of various factors among grouped subjects**

Study subjects (n=33)	Whole group (n=33)	Group I CDH (n=18)	Group II CDH+sac (n=6)	Group III DE (n=7)	Group IV- Paraesophageal and morgagni hernia (n=2)	P	Significance
Age at presentation (days), mean±SD	271±399.12	244±353.8	403±624.5	270±374.8	112.5±95.46	0.804285	NS
Sex, n (%)							
Male	19 (57.57)	11 (33.33)	3 (9.09)	4 (12.12)	1 (3.03)	0.030202	S
Female	14 (42.42)	7 (21.21)	3 (9.09)	3 (9.09)	1 (3.03)		
Male:female ratio	1:0.74	1:0.64	1:1	1:0.75	1:1		
MOD, n (%)							
Preterm	2 (6.06)	1	1	0	0		
NVD	1 (3.03)	1 (3.03)	0				
LSCS	1 (3.03)	0	1 (3.03)				
NVD:LSCS ratio	1:1	1:0	1:0	0:0	0:0		
Full term	31 (93.93)	16 (48.48)	6 (18.18)	7 (21.21)	2 (6.06)	0.049282	S
NVD	19 (57.57)	8 (24.24)	4 (12.12)	5 (15.15)	2 (6.06)		
LSCS	12 (36.36)	8 (24.24)	2 (6.06)	2 (6.06)			
NVD:LSCS ratio	1:0.63	1:1	1:0.5	1:0.4	1:0		
Gestational age weeks, mean±SD	38.54±1.71557	38.39±1.79	37.83±1.33	39.42±1.81	39±1.414	0.378637	NS
Associated congenital anomalies, n (%)	6 (18.18)	2 (11.11)	2 (33.33)	1 (14.29)	1 (50)		
ARM	2 (12.12)	1 (5.55)	1 (16.16)	0	0		
Cardiac	4 (6.06)	1 (5.55)	1 (16.16)	1 (14.28)	1 (50) (OSASD)		
Others	0	0	0	0	0		
Birth weight (kg)	2.74±0.573504	2.65±0.51	2.78±0.53	2.5±0.79	3.25±0.3535	0.55076	NS
Surgery weight (kg)	4.742±2.451	4.54±2.97	5.04±2.25	5.09±2.57	4.73±1.796	0.945741	NS
Pre-operative ventilation, n (%)	3 (9.09)	3 (9.09)	0	0	0		
Post-operative ventilation, n (%)	3 (9.09)	3 (9.09)	0	0	0		
MOS, n (%)	32 (96.96)*	17/32 (53.12)*	6 (18.75)*	7 (21.88)*	2 (6.25)*	0.168	
Open, n (%)		10/17 (58.82)	2/6 (33.33)	2/7 (28.58)	1 (50)		
Thoraco, n (%)		5/17 (29.41)	4/6 (66.66)	5/7 (71.42)	0 (0)		
Lapar, n (%)		0 (0)	0 (0)	0 (0)	0 (0)		
TAO, n (%)		1/17 (5.88)	0 (0)	0 (0)	0 (0)		
LAO, n (%)		1/17 (5.88)	0 (0)	0 (0)	1 (50)		
DOHS (days), mean±SD	16.27±10.05694	16.89±11.68	14±8.84	18±8	11±5.657	0.796622	NS
Reccurrence, n (%)	0	0	0	0	0	0	
SSI, n (%)	0	0	0	0	0	0	
Other surgeries, n (%)	2 (6.25)	1/17 (5.88) (ARM-3)	1 (16.66) (ARM-3)	0	0	0	
Survival, n (%)	31 (93.93)	15/17 (88.24)	6 (100)	7 (100)	2 (100)	100%	

NS: Not significant, S: Significant, SD: Standard deviation, CDH: Congenital diaphragmatic hernia, LSCS: Lower segment caesarean section, NVD: Normal vaginal delivery, MOD: Mode of delivery, SSI: Surgical site infection, ASD: Anterior septal defect, OSASD: Ostium secundum ASD, PDA: Patent ductus arteriosus, ARM: Anorectal malformations, MOS: Mode of surgery, TAO: Thoracoscopic assisted open, LAO: Laparoscopic assisted Open, DOHS: Duration of hospital stay

There were only 2 (6.06%) preterm deliveries both delivered by caesarean section in Group I in the study, otherwise all were full-term deliveries: 8 born by full term normal vaginal delivery (FTNVD) (24.24%) and 8 by lower segment caesarean section 8 (24.24%) in Group I and 6 (18.18%) in Group II.

The mean gestational age in weeks in Group I was 38.39 ± 1.79 (median: 38, range: 37–40), Group II had 37.83 ± 1.33 (median: 37.5, range: 36–40), Group III had 39.42 ± 1.81 (median: 40, range: 37–42 weeks) and in Group IV, the mean was 39 ± 1.41 weeks.

The mean birth weight in Group I was  $2.65 \pm 0.51$  (median: 2.5, range: 1.75–3.8.), in Group II, it was  $2.78 \pm 0.53$  (median: 2.5, range: 2.3–3.75 kg.), in Group III, it was  $2.5 \pm 0.79$  (median: 2.5 range: 1.7–4.1), whereas Group IV had a mean of  $3.25 \pm 0.3535$ .

The mean weight at the time of surgery in Group I was  $4.54 \pm 2.97$  (median: 2.5, range: 1.9–3) Group II had  $5.04 \pm 2.25$  (median: 4.5, range: 1.9–3), in Group III, it was  $5.09 \pm 2.57$  (median: 3.5, range: 2.36–9.75) and in Group IV, the mean was  $4.73 \pm 1.796$ , which was not significant statistically.

Only 3 neonates (9.09%) required pre-operative ventilation for average of 3 days in Group I due to PPHN, none in other groups. One baby required high-frequency ventilation for 5 days.

## THE DISCUSSION

The diaphragm is a large, dome-shaped musculotendinous structure separating the pleural and peritoneal cavities. Embryologically, the diaphragm develops from septum transversum of the mesoderm. The defect in CDH results from failure of the pleuroperitoneal canals (effected by growth of the posthepatic mesenchymal plate and of the pleuroperitoneal folds) to close at the end of the embryonic period (8<sup>th</sup> gestational week), resulting in the gut entering the thoracic cavity through this defect eventually causing compression and finally hypoplasia of the affected lung.<sup>[1,2]</sup>

The left posterolateral diaphragm closes after the right side, which explains the majority of Bochdalek hernias occurring on the left side containing spleen, stomach small intestine and colon, whereas right-sided hernias are much rare and containing liver, gallbladder and intestines.

Multiple genetic factors along with environmental and nutritional factors have been proposed to be the possible aetiologies for CDH.<sup>[1,2]</sup>

The present study was a combination of retrospective and prospective observational cohort study of right-sided CDH in children including in neonates to measure risk factors for mortality and outcomes of right CDH (RCDH). CDH has been recognised as a syndrome, which includes pulmonary hypoplasia, lung immaturity, left heart hypoplasia and PPHN of the newborn.

The prenatal detection rate for CDH varies enormously in published studies, from 10% to 79%. Most are detected after 24 weeks of gestation. Few studies have quoted as earlier detection is associated with increased mortality. However, prenatal detection of CDH is rare in developing countries due to inadequate facilities. In our cohort, 5/33 (15.15%) of the cases were diagnosed prenatally; most were detected at 32–35 weeks, just before delivery. Polyhydramnios has been reported 10/33 (30.30%) cases.

Literature says antenatal detection rate of RCDH is less compared to left CDH (LCDH).<sup>[1,2]</sup>

Most commonly CDH is diagnosed immediately after birth with respiratory distress, scaphoid abdomen, bowel sounds in the chest and mediastinal shift on chest X-ray. However, in exceptional cases, it can present with chronic respiratory issues like recurrent bronchopneumonia.

Surgical repair of CDH is the treatment of choice and should be undertaken only after cardiorespiratory functions are stabilised. A policy of ‘delayed’ surgery coupled with gentle ventilation and occasionally extracorporeal membrane oxygenation (ECMO) support yields the best results recorded.

Preoperatively, all of our babies were examined for associated malformations. We had 8/33 (18.18%) babies with associated malformations, 4 (9.09%) each of anorectal malformation (ARM) and cardiac non-lethal malformations. ARM babies included anterior ectopic anus, rectovestibular fistula, rectobulbar fistula and rectovaginal fistula, each requiring staged procedure.

Cardiac malformations are most common anomalies associated with CDH; however, their aetiology is still unclear. None of our babies had cyanotic congenital heart disease/lethal or any associated syndromes, including atrial septal defect, ventricular septal defect and patent foramen ovale.

Despite the progress in prenatal diagnosis and intervention as well as postnatal therapeutic strategies, CDH is still associated with significant mortality because of the induced pulmonary hypoplasia.

Most of cases were diagnosed by routine chest X-ray and ultrasonography. Only in doubtful cases, contrast-enhanced chest tomography and very rarely upper gastrointestinal contrast study were done.

Repair of the RCDH may be accomplished through an open or minimally invasive approach: either by open laparotomy/thoracotomy or thoracoscopy/laparoscopic approaches. Regardless of the mode of the approach, the tensionless, proper closure of the defect was critical [Figures 1a-d and 2].

Under general anaesthesia, a right sub costal or transverse abdominal incision is made, the herniated liver was gently reduced into the abdomen and the diaphragmatic orifice is closed with interrupted non-absorbable sutures. Placement of inter costal drainage tube was left to surgeon’s decision [Figure 1a-d].

It has been speculated that partial liver herniation, which is a common finding in the RCDH, may block the further herniation of hollow viscera, preventing the development of gastrointestinal symptoms.<sup>[1,2]</sup>

There has been an increasing trend towards thoracoscopic repairs, which are thought to minimise post-operative pain and scarring and hasten recovery.<sup>[1-3]</sup>

Stable babies were tried minimal access surgery thoracoscopic or laparoscopic placcation; however, in technically difficult situations or in medical instability, they were converted to open procedures. Medically unstable babies were tried directly open

reduction and repair. The overall survival rate in our study was 93.93% [Figure 2]. Several series have demonstrated higher recurrences rates following thoracoscopic approach, although these may have been biased by higher rates of unfavourable anatomy and an inherent learning curve.<sup>[3]</sup>

32/33 (96.96%) babies underwent surgical repair under general anaesthesia. 17/32 (51.51%) in Group I, 6 in Group II, 7 in Group III and 2 in Group IV underwent surgical repair successfully. One neonate expired preoperatively (1/33, 3.03%) due to PPHN.

5/32 (15.15%) in Group I, 4/6 (12.12%) in Group II and 5/7 (12.12%) in Group III underwent thoracoscopic repair. One (3.03%) each in Group IV and Group I underwent laparoscopic converted to open repair due to technical and medical reasons; rest all babies underwent open laparotomy repair.

Babies with diaphragmatic agenesis uniformly require placement of a patch to close the diaphragmatic defect. In our study, we did not encounter such babies; hence, mesh repair was not done in any of our babies.

The overall lower incidence of RCDH has limited our understanding of the disease. Prognostic indicators for survival applied to LCDH have been extrapolated and have been used as survival indicators for the RCDH, which is practically not accurate for clinical as well as statistical purposes.

Mortality rates varied from 3% to 32% as per the limited literature available.<sup>[1-3]</sup>

Cohen-Katan *et al.* in their study found that right-sided CDH carried higher mortality compared to LCDH with an incidence of 81.25%; in our study, we had mortality rate of 6.25%.<sup>[4]</sup>

Having good birth weight  $2.74 \pm 0.57350$  as well as surgical weight  $4.742 \pm 2.451$  in our study explains overall good prognosis of referred babies in our centre; however, both were not significant statistically. This highlights the importance of early suspicion of the diagnosis and early hence early referral.

2/31 were premature in Group I, one born by normal, another by caesarean section with a ratio of 1:1. The rest of all babies (31/33, 93.93%) were born full-term deliverers, 19 were normal deliveries and 12 were born by caesarean section, with a ratio of 1:0.63. Both were significant statistically.

In our study, we found sex ratio and mode of delivery statistically significant.

Overall survival was 93.93% in our study with 15(83.33%) in Group I, 6(100%) in Group II, 7(100%) in Group III and 2(100%) in Group IV. Two babies in Group I could not be saved due to unstable medical conditions and due to persisting primary pulmonary hypertension.

Previous studies have shown that the recurrence rate after thoracoscopic surgery is from 5.0% to 25% when compared to open repair.<sup>[3]</sup> However, our study showed that endoscopic repair of RCDH is safe, effective and the clinical efficacies

were as comparable with those of open surgery. With the application of the endoscopy technology and a gain in surgical skills, endoscopic repairs of right diaphragmatic defects could be the preferred method by which to treat them.<sup>[5]</sup>

Tiryaki *et al.* stated from their study the incidence of higher recurrence in a thoracoscopic approach.<sup>[6]</sup>

The presence of a hernial sac significantly improves the prognosis in CDH neonates which is formed of parietal peritoneum and lung pleura and has been reported in approximately 20% of cases in the literature. In our study, we found intra operatively, the presence of sac in 6/32 (18.75%) neonates; all of them survived, wherein histopathological examination was done proved to be devoid of muscle fibres.<sup>[7]</sup>

Two babies 2/33, one in each in Group I and Group II, underwent ARM corrections in stages apart from CDH repair successfully.

Three babies (3/32, 9.09%) in Group I did require pre- and post-operative ventilatory support. Since in our institute ECMO facility is not available, one baby required high-frequency oscillatory ventilation postoperatively.

The duration of hospital stay (in days) in our study was  $16.27 \pm 10.0569$ , which was not significant statistically.

In our study, we did not encounter any surgical site infections or recurrences, with exceptionally excellent survival rates postoperatively.

In 1769, Giovanni Battista Morgagni, an Italian anatomist, described an anterior retrosternal diaphragmatic defect occurring between the xiphoid process of the sternum and the costochondral attachments of the diaphragm and constitutes <2% of reported diaphragmatic defects.<sup>[8-11]</sup> We had one case of right-sided Morgagni hernia which was managed by the laparoscopic approach which was converted to open laparotomy to close the defect due to technical reasons. We had one baby with right para-oesophageal hernia presenting with bronchopneumonia repaired by laparotomy successfully [Table 1].

Although ours was a short-term study with small number of referred cases with excellent survival rate, long-term follow-up is however required to know the prognosis of pulmonary as well as other comorbidities [Figure 3].

The limitations of this study were that antenatal ultrasonographic findings (lung head ratio, liver up and stomach up), OI's values and Apgar scores were not consistently available in the medical case records since ours is referral centre.

## CONCLUSION

Right-sided congenital diaphragmatic defects, though rare, do carry excellent survival if referred early and managed in a tertiary care neonatal and paediatric centre as that of left diaphragmatic defects. With the use of available conventional

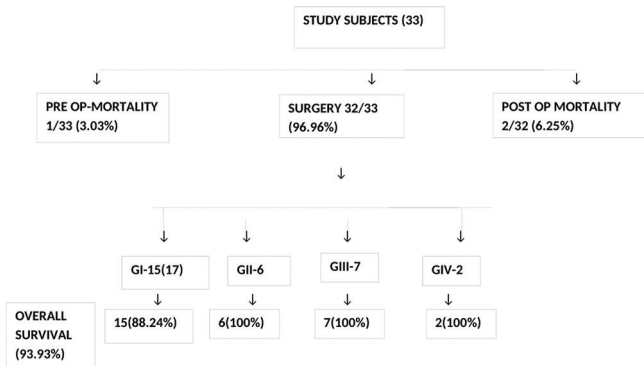


Figure 3: Consort diagram

ventilatory facility, excellent survival results can be achieved with dedicated team efforts.

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**Conflicts of interest**

There are no conflicts of interest.

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