Research Article

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Pre-Operative Diagnosis of Infantile Triad of Waugh's Syndrome Associated with Hypoganglionosis-Key Radiological Findings

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Abstract

Intestinal malrotation and intussusception each carry a significant mortality risk and are regularly seen by clinicians treating paediatric emergencies. Waugh's syndrome is a combination of intestinal malrotation and intussusception which is rare and has only been reported as intra-operative findings in surgical literature. Hypoganglionosis (HG) is a rare condition that is characterised by a reduced number of ganglion cells in the submucosal and myenteric plexuses of the colon and thickened muscularis mucosae and muscularis propria layers. There are considerable radiological challenges and clues to diagnosis and subsequent treatment in this particular triad combination of life-threatening conditions. We describe a presentation of Waugh's syndrome and hypoganglionosis to report a new association and highlight specific radiological challenges for all clinicians treating a similar case. A male infant having diagnosed hypoganglionosis who had constipation since early infancy and treated conservatively developed Waugh's syndrome which was diagnosed prospectively. Clinical features, plain abdominal radiograph, ultrasound with color Doppler, negative air enema contrast films and most importantly our previous experience were helpful in diagnosing the condition. Attempts at repeated air enema reduction of intussusception failed and at exploratory laparotomy intussusception was reduced manually with correction of malrotation by Ladd's procedure without an incidental appendicectomy and the patient recovered well. We aim to highlight the diagnostic challenges and radiological clues in such cases and report a new association between Waugh's syndrome and hypoganglionosis.

Keywords: Acute Abdomen; Hypoganglionosis; Imaging; Intussusception; Malrotation; Radiological; Waugh's Syndrome

Introduction

We previously evaluated and recently published the various triad lesions instead of dual pathology routinely described with Waugh's syndrome [1]. Pediatric triad with Waugh Syndrome was first reported in 2014 and Infantile Waugh's syndrome is even rarer [2,3]. Waugh's syndrome is the association of intussusception and intestinal malrotation first named by Brereton et al in 1986[4]. Its pre-operative diagnosis is rare unless looked for actively and we report a case in which high index of suspicion in combination with clinical, radiological, imaging, interventional procedures and our previous experience with this condition helped to establish the diagnosis and subsequently had undergone successful treatment. We describe a rare case of Infantile Waugh's Syndrome, which is a combination of intussusception and intestinal malrotation in association of hypoganglionosis and the importance of a high index of suspicion and the use of various diagnostic tools and procedures in establishing a diagnosis.

Case Report

A 6-month-old boy presented with 12-hour history of intermittent crying, vomiting, constipation and irritability. Patient was first born baby at term with normal vaginal delivery and had passed meconium and urine soon after birth. The baby had no constipation in neonatal period. The baby developed chronic constipation and abdominal distention in early infancy for which plain abdominal radiograph showed rectosigmoid dilatation with a mild transition zone like tapering at rectosigmoid junction and reversed rectosigmoid ratio which was confirmed with ultrasound with incompletely filled bladder. With persistent symptoms needed to rule out the Hirschsprung's disease or allied Hirschsprung's disorders or variant Hirschsprung's disease like congenital colorectal secreto-motility challenging disorders requiring various stains and histochemical technology. A full thickness rectal biopsy showed presence of ganglion cells, no hypertrophic nerves or increased acetyl cholinesterase enzymes and ruled out HD. However, it showed absence of acetylcholinesterase in the mucosa, reduction of nerve cells in the myenteric and submucosal plexus and reduction in the number of ganglion cells per longitudinal centimeter was decreased by a factor of 2, and the development of nerve fibers was scarce thus established the diagnosis of hypoganglionosis. The parents were unwilling to undergo operative correction and was treated with conservative therapy which consisted of enema therapy initially followed by suppositories as required and Movicol pediatric laxative therapy to reduce the resistance and evacuate the dilated colorectum to come back to normal and reduce the risk of ileocecal reflux and insulating the subsequent small bowel intestinal bacterial overgrowth, micronutrient supplements, increasing prebiotic, probiotic, postbiotic and holobiotic therapy, and encouraging to take more fluids and fibers in the diet and continue breast feeding as much as possible. The patient improved, all symptoms disappeared, second option with surgeons not familiar with this kind of new disorders did not support it and hence parents have stopped for the past few weeks to test the second opinion was correct or not.

On examination. a palpable mobile, mildly tender, sausage shaped smooth 3-4 cm diameter mass in the left paramedian region was left.

Laboratory investigations were normal. A plain abdominal radiograph depicted an abnormal bowel gas pattern in the form of featureless bowel loops in the central and upper abdomen with a cut-off in the left hypochondrium. and gasless flanks and pelvis suggestive of features of intestinal obstruction with a possibility of an intussusception (Figure 1). Ultrasound scan with color Doppler confirmed the diagnosis with doughnut and pseudo kidney signs. At the level of the splenic flexure of the colon, there was a typical target round image of intussusception which has weak Doppler signal within (Figure 2). The patient was resuscitated with a bolus of 20ml/kg of normal saline followed by maintenance fluids and intravenous co-amoxiclav antibiotic.

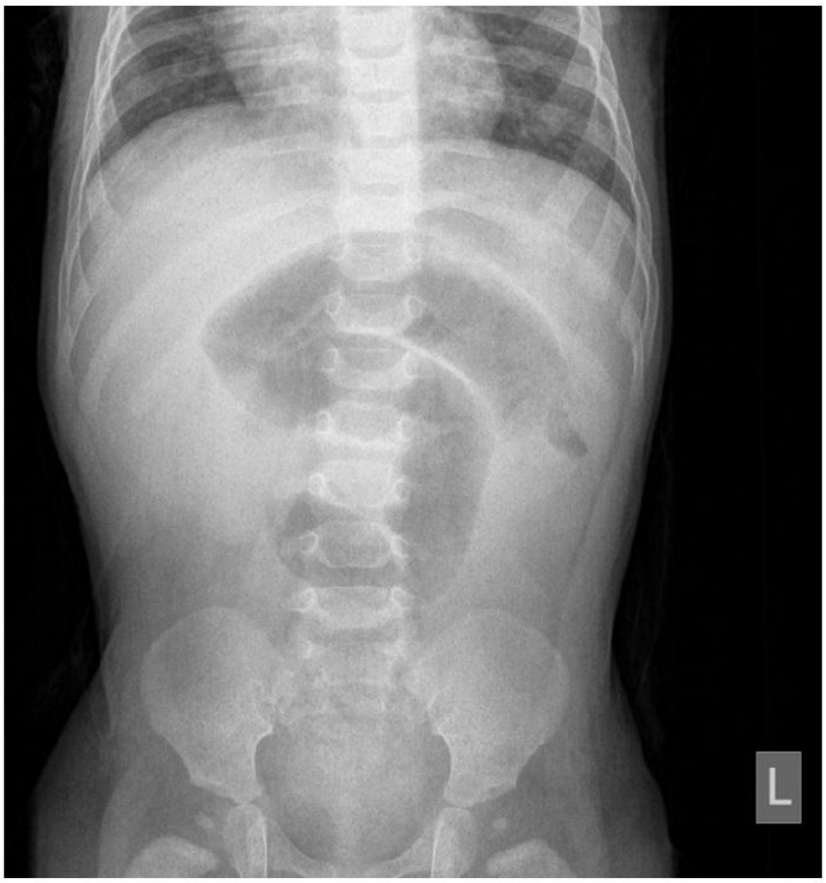

Figure 1: Abdominal radiograph with abnormal central bowel gas and empty pelvis and flanks.

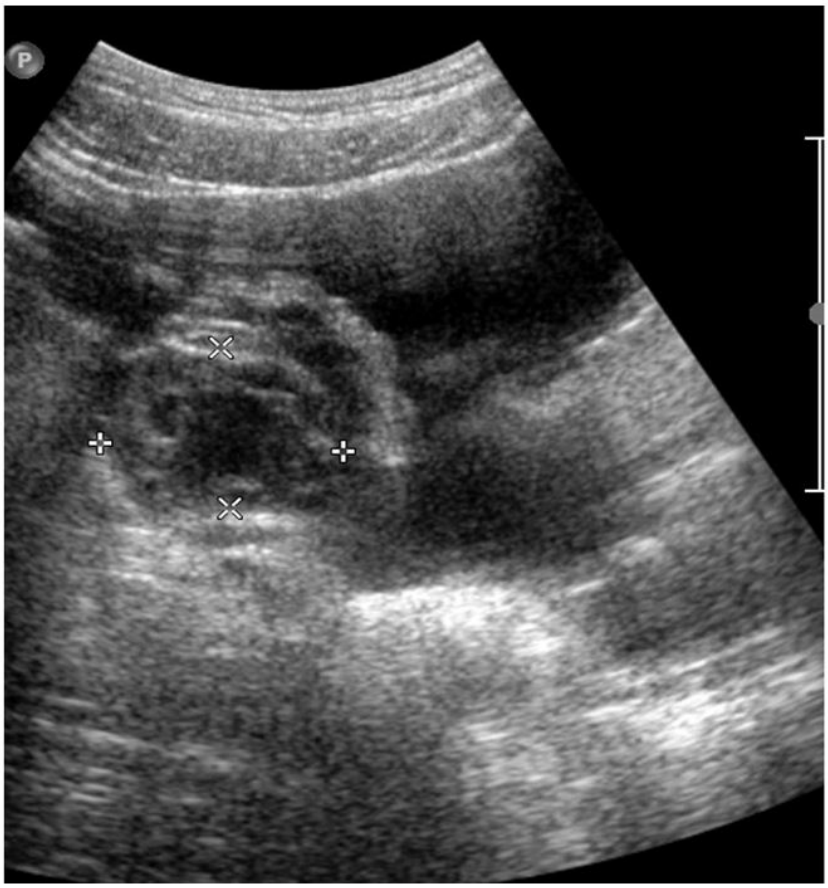

Figure 2: Typical target sign near splenic flexure area on ultrasound.

A therapeutic air enema reduction was immediately attempted. Under fluoroscopy the rectal huge dilatation with fecal loading was very much evident with reversed rectosigmoid ratio and clear transition zone at the rectosigmoid junction. The left colon was tight with resistant to open up with disordered waves of peristalsis and maximum resistance was felt at the junction of midgut and hind gut at the left transverse colon where there is a physiological transition with a small functional sphincteric mechanism. Air filled the colon in a retrograde manner, beyond the splenic flexure and into the right colon allowing pneumatic dilation of the left colon thereby reducing the distal resistance. It was not possible to produce any further stepwise luminal filling of air and the procedure was terminated. Subsequent ultrasonography now demonstrated the target appearance of an intussusceptum in the right upper quadrant and a second therapeutic air enema was performed. At the air enema reduction, an intussusception was at the splenic flexure of the colon and stepped back progressively until at the level of the ileo-cecal valve which was abnormally located in the subhepatic region, although it stopped progressing at right upper quadrant and after two attempts the procedure was stopped [Figure 3]. A new third attempt at reduction was performed without success.

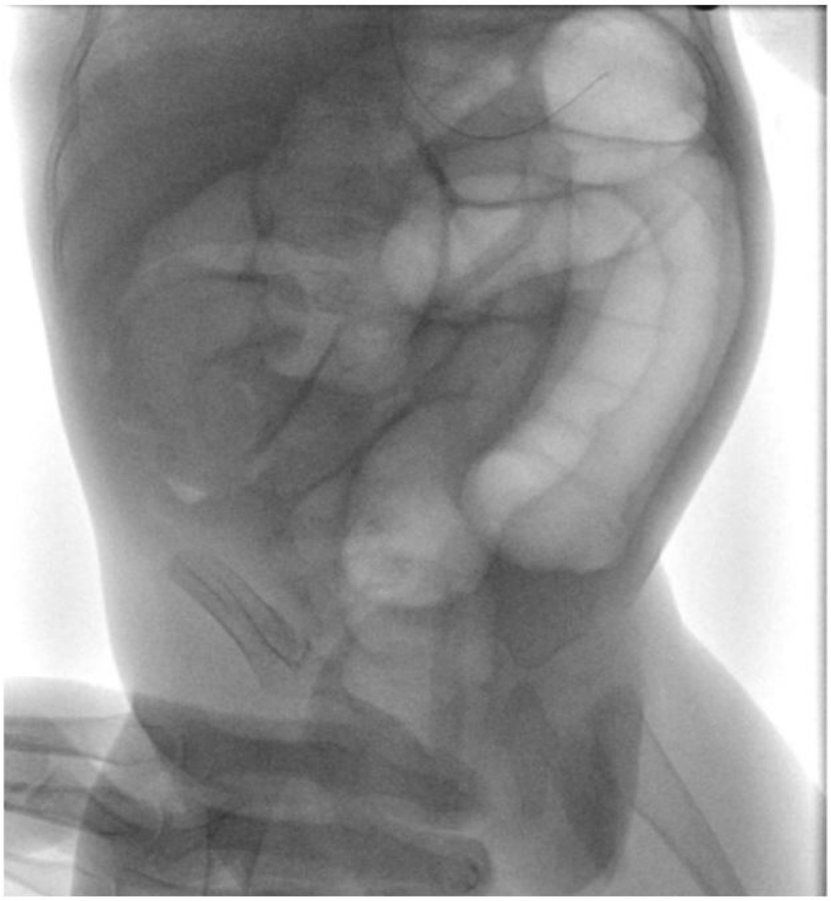

Figure 3: Air enema reduction fluoroscopy film with intussusception in the subhepatic area after 3 attempts.

An abdominal radiograph at this stage showed cecum in right upper quadrant and prominent haustra on the upper and left side of the abdomen with characterless bowel loops in the right lower quadrant and central abdomen with transition zone in negative contrast at the rectosigmoid junction with collapsed rectum and dilated left and transverse colon due to underlying hypoganglionosis suggesting irreducible intussusception with malrotation. Midgut malrotation with intussusception (Waugh's syndrome) was considered [Figure 4].

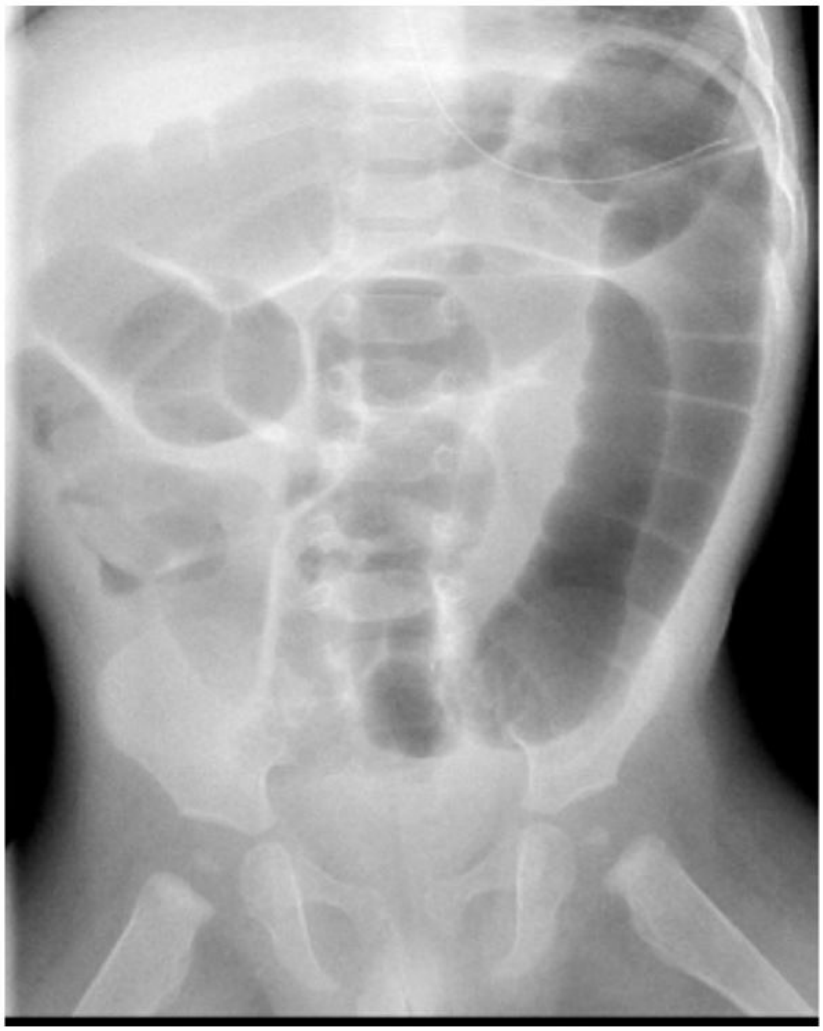

Figure 4: Post-air enema reduction abdominal radiograph. Note transition zone in negative contrast at the rectosigmoid junction with collapsed rectum and dilated left and transverse colon due to underlying hypoganglionosis.

The radiological features of hypoganglionosis during fluoroscopy and radiographs at air enema reduction included a transition zone at rectosigmoid junction, abnormal rectosigmoid index, dilated left and transverse colon, and retention of air on delayed film. In addition, irregular hind gut contraction, lengthening of the colon, and redundancy were observed.

The patient underwent exploratory mini-laparotomy via a small periumbilical incision, reduction of ileo-colic intussusception, confirmation of midgut malrotation and its correction by Ladd's procedure. The Ladd's procedure is popularized by Dr William Ladd, the father of American and global pediatric surgery which consist of dividing the compressing fibrous bands stretching between the gall bladder and cecum across the second part of duodenum causing partial external obstruction which is known as Ladd's bands, widening the base of the small intestinal mesentery by placing the duodenojejunal junction well to the right of the midline and moving the ileocecal junction to the left upper quadrant in the place of left splenic flexure region as in midgut malrotation the duodenojejunal flexure is to the right of the midline making C of the duodenal loop into an L shape and the ileocecal junction is subhepatic just seating over the duodenojejunal flexure making zero length of the small bowel mesentery and predisposing the pedicle of combined proximal and distal junctions to volve around the single axis easily and develop catastrophic midgut volvulus which if not recognized early may end in the loss of almost all small bowel and right colon except the first and second part of the duodenum and colon beyond mid transverse region. Incidental inversion appendectomy was avoided, for various reasons explained in the discussion, at the end of the procedure Patient recovered uneventfully. At 12 month follow up he was asymptomatic, thriving well.

Discussion

Although it was called Waugh syndrome in 1986, it was first described in 1911 by Waugh [4]. A rarely reported entity, Waugh's syndrome is the association between intestinal malrotation

and intussusception [1-12]. To the best of our knowledge an association of hypoganglionosis with this syndrome with possible implications on its causation and pre-operative diagnosis has not been reported. Our case emphasizes the rarity of infantile Waugh syndrome, and the importance of being aware of its potential symptoms and presentations to aid in early diagnosis as the existence of one congenital pathology of hypoganglionosis should alert to the possibility of other associated anomalies of the midgut malrotation which in turn predisposes to intussusception formation. Bilious vomiting in infants should alert to the possibility of underlying malrotation as well.

Midgut malrotation by its nature is associated with a mobile right colon, which may be a prerequisite for intussusception [8]. Moreover, the angle of ileo-caecal junction is more obtuse and telescoping becomes much easier. The association may be more common than recognized as non-operative reduction of intussusception may fail to recognize the associated malrotation as opposed to open procedures in which it has been found in 40% of cases [1].

The association is more frequent in infants but has been reported in adults as well. Prolonged pre-operative intermittent symptoms should alert the clinician to suspect underlying midgut malrotation especially in cases of associated other congenital anomalies as was the case with our patient. Moreover, abdominal mass on the left periumbilical region even in a recently started case should raise further suspicion as most initial cases should have lump in the right periumbilical area.

Plain abdominal radiograph can give indication of the bowel gas pattern abnormalities raising suspicion for associated malrotation. The air enema may provide negative contrast studies and the colon occupying left side of the abdomen with caecum lying high and very mobile gives further clues. Quite often non-operative treatment fails and may have higher incidence of recurrence if the underlying malrotation which predisposes it is not corrected at the same time.

The key radiological features are the hyper mobile right colon demonstrated on a timeline of ultrasound and fluoroscopic examinations. Due to the constant axial positional change of the intussusceptum, from the right lower to left upper quadrant, the radiologist is challenged to recognize the possibility of malrotation, whilst performing the therapeutic challenge of reducing the intussusception. This hyper mobility has been a previous surgical finding and may in fact predispose to intussusception[4]. There may be few or no other clues to suggest abnormal gut lie. As ultrasound is used to identify intussusception, the relative orientations of the superior mesenteric artery and vein could be used to screen for abnormal rotation in inconclusive cases [1,3]

Once diagnosed, most cases will need exploratory laparotomy but recently laparoscopic approach has been applied successfully. Operative reduction is successful in most cases but some advanced cases may need resection [9-10]. Pre-operative diagnosis of this triad following key radiological findings combined with high index of suspicion based on our past experience, allowed us to plan a periumbilical minimal invasive option as opposed to right upper transverse or right lower quadrant approach to avoid confusion with appendectomy later in life.

Incidental appendicectomy is usually an integral part of the Ladd's procedure which is carried out as inversion appendectomy after dividing the mesoappendix and inverting the appendix into the cecum to avoid opening the lumen and converting a clean procedure into contaminated one to avoid postoperative sepsis and adhesions. It was avoided as part of the Ladd's procedure in our case for several reasons. The incidence of recurrent and post-operative intussusception is more common in this association and may make air enema reduction more hazardous simply by blowing off the appendicular stump causing pneumoperitoneum and subsequent peritonitis. Secondly the inverted appendix may form a lead point and induce an intussusception later on. Finally, the appendix being tonsil of the tummy, we believe that appendix is an equivalent of the bursa of Fabricius in avian species responsible for the differentiation and production of local antibody formation by B lymphocytes, like pharyngeal tonsil being very important in young children to develop sound immunity we try to avoid appendectomy at least in the first decade if possible and feasible. However, not performing appendicectomy with Ladd's procedure may make diagnosis of acute appendicitis more difficult at a later date due to abnormal position. Increasing use of laparoscopy should overcome this difficulty.

Hypoganglionosis, being congenital, has higher chances of midgut malrotation as it involves the hind gut and secondarily can cause effects on the midgut duodeno-jejunal flexure leading to incomplete rotation predisposing for malrotation of the midgut in the first instance. Intestinal malrotation may predispose patient to intussusception due to obtuse rather than right angle at the ileo-cecal junction. Hypoganglionosis has its own radiological findings at different ages in the neonates, infants, children and later in adult life [13]. The association of hypoganglionosis with malrotation may predispose the development of midgut malrotation which can predispose the development of the intussusception due to ileocecal obtuse angle. Based on the evidence of experimental fetal gastroschisis model in fetal rabbits, we believe that the congenital hypoganglionosis of the rectosigmoid and left colon may predispose the element of incomplete rotation or a malrotation of the midgut due to blind loop obstruction created by a competent ileocecal valve

leading to raising the ileocecal junction higher up and push the duodenojejunal flexure to the right due to pressure exerted by junction of midgut and hindgut at left transverse colon region finally leading to merging both junction superimposed on each other[14].

Conclusion

In conclusion we believe that incidence of malrotation should be suspected in cases with other congenital anomalies like hypoganglionosis as was the case in our patient. The association of malrotation and intussusception is more frequent than recognized and chances of non-operative reduction or post reduction recurrence are higher in this group of patients. As we have demonstrated, a pre-operative diagnosis based on key radiological findings of this Infantile triad has distinct advantages as forewarned is forearmed with adequate preparation and armamentarium to correct these problems with special attention. Air enema reduction of intussusception may be inadequate if a malrotation component is present. In the post reduction period, midgut malrotation associated volvulus could be misdiagnosed as recurrent intussusception. As the Ladd's procedure predisposes to post-operative intussusception, avoid appendicectomy which otherwise precludes air enema reduction due to fear of stump blow out but appendicitis later in life may be difficult to diagnose. Highlighting the importance of a high index of suspicion and the use of clinical, radiological, and interventional techniques in making a diagnosis of Waugh's syndrome could be useful for patients, parents, public and, other healthcare professionals alike.

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Author Contributions

All authors contributed to the study conception and design. Material preparation, data collection and analysis were performed by all authors. The first draft of the manuscript was written by RVP, and all authors commented on or edited previous versions of the manuscript. All authors read and approved the final manuscript.

Compliance with Ethical standards

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Conflict of Interest

The authors declare that there is no conflict of interests that prejudices the impartiality of this scientific work.

Informed Consent

Informed consent was obtained from the parents and all the relatives involved prior to all the procedures. Parents and all involved parties were informed about the procedure.

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