

# E-POSTERS

## Abstracts



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## GOLDENHAR SYNDROME: A RARE CASE REPORT

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**Background:** Goldenhar syndrome, or oculo-auriculo-vertebral syndrome, is a poly-malformative syndrome associating most often an epibulbar dermoid cyst, pretragial diverticula and vertebral anomalies. We report a rare observation of Goldenhar syndrome associating lateral labial cleft, dermoids of the ocular limbus and pre-tragic excretion

**Materials & Methods:** This is the observation of female child 2 years old presenting a Goldenhar syndrome associating lateral cleft lip, dermoids of the ocular limb and pre-tragic excretion and pre-auricular diverticula. Complimentary examinations, namely stromal radiograph of the echocardiography spine, did not show any associated abnormalities. The hospital where the surgery has taken is : EHS El-Mensourah Constantine, Child and Adolescent Hospital, Constantine, Algeria

**Discussion:** Goldenhar syndrome is a rare malformation syndrome related with an anomaly of development of the first gill arches. Although most cases of SDEG are sporadic, the modalities of autosomal recessive inheritance and the dominant have been described. Also referred to as oculo-atrial dysplasia, it is most often unilateral affecting the soft tissues and less the bone component. It usually includes peribulbar dermoids or dermolipomas, pre-auricular cuticular diverticula, abnormalities of the shape of the auricle, conductive hypo-acoustics; abnormalities of the facial area and sometimes vertebral anomalies. It can be added to these malformative alterations a coloboma of the eyelid, a Duane syndrome and very rarely microphthalmia or anophthalmia.

**Conclusion:** Through this observation, we discuss the pathogenic aspects and clinics of this rare clinical entity and the therapeutic measures taken.

### Biography

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She is board-certified in surgery and pediatric surgery, and has dedicated interest in pediatric urology and tumors, as well as neuroblastoma, childhood sarcomas. She also enjoys general pediatric surgery including plastic and neonatal care. She completed her medical degree at Constantine Algeria Medical School, and her general pediatric surgery residency at the EHS El Mensourah Constantine Hospital.

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# SKELETAL MUSCLE METASTASIS: AN UNUSUAL MANIFESTATION OF ADENOCARCINOMA OF LUNG

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**Introduction:** Lung carcinoma, the most common malignancy worldwide, presents as a metastatic disease in majority of the cases. The most frequent sites of distant metastases are liver, adrenal glands, bones, and brain. Skeletal muscle metastasis is an unusual presentation of lung adenocarcinoma.

**Case report:** A 53 year old male patient, labourer by occupation, *beedi* smoker since 25 years, admitted to tertiary care hospital with pain and swelling over left arm, cough and expectoration since 2 months, accompanied with weight loss of three kilograms. There was no evidence of chest pain or hemoptysis. On examination, there was hard swelling over extensor compartment of left upper limb with mild tenderness, no loss of sensation and mild restriction of range of movements on flexion at elbow. Respiratory and other system examination was within normal results. CT chest pain defined multilobulated lesion in right perihilar location in right middle lobe. USG study of left arm showed an irregular heterogenous soft tissue lesion noted within the triceps muscle with few areas of intra-lesional necrosis, MRI of left arm showed lobulated lesion in posteromedial aspect of mid and distal arm, involving triceps muscle, medial aspect of brachialis, encasing brachial artery, veins and median nerve. PET CT showed enhancing nodular soft tissue lesion noted in middle lobe of right lung, 2.9x2.4 cm. Biopsy revealed metastatic adenocarcinoma. For further studies immunophenotyping was done which showed negative for EGFR and ALK. Patient was treated with palliative RT, Pem-Carbo f/b pemetrexed maintenance and recently 3# of Gemcitabine. The patient died of metastasis to brain within eight weeks of diagnosis.

**Conclusion:** Lung carcinoma with skeletal muscle metastasis should be considered as a potential differential diagnosis in patients presenting with intramuscular mass.

## Biography

Sharan Badiger has completed his MD in Internal Medicine, in 1994 and MBBS, in 1989 from Gulbarga University, Gulbarga, Karnataka, India. He is presently working as Professor of Medicine, Sri B M Patil Medical College, Vijayapur, Karnataka, India. His main research interests are in Internal Medicine, Echocardiography and Imaging in Medicine. He has 60 publications in international/national reputed journals and proceedings of the conferences. He is a Life Member of Association of Physicians of India since 1996, Research Society for the Study of Diabetes in India since 2005, Indian Society of Cardiology since 2006, Indian Society of Electrocardiology since 2010, Indian Academy of Geriatrics since 2010, International Association of Computer Science and Information Technology Singapore since 2010. He is an international Associate Member of American College of Cardiology since 2014. He is serving as Editor-in-Chief, Editorial Board Member, Peer Reviewer and Advisory Board Member of various national, international journals and conferences of repute.

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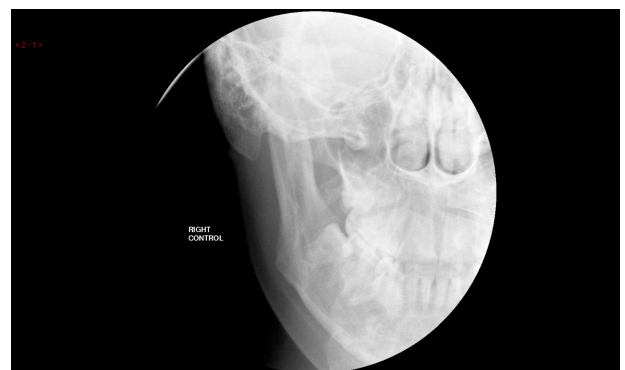
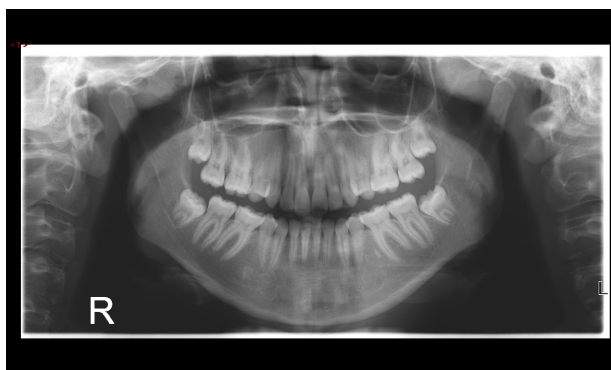
# RIGHT-SIDED FACIAL SWELLING WITH FUNCTIONAL HEMIPARESIS: A DIAGNOSIS DILEMMA

**Claire Graham, Samantha Reid, Colette Balmer and Kathryn Taylor**

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**F**unctional paresis is a psychogenic phenomenon resulting in patients complaining of weakness and disturbances in function without pathology. Often patients with function symptoms have a history of depression and anxiety. Here we discuss a case of an 18 year old female who self-referred into the local dental emergency clinic regarding an acute right-sided facial swelling and a right-sided facial paresis. Further investigation revealed the likely cause of her right parotid swelling was a mucous plug or sialolith that subsequently passed. It transpired the right facial paresis which was not a new presentation and the patient had been under the care of a consultant neurologist for functional hemiparesis. Interestingly, the patient had worked in a stroke ward at the time of onset of the hemiparesis. Due to the limited history expressed by the patient and clinical findings, upper motor neurone pathology was suspected initially. This case highlights the importance of liaising with other specialties in order to proceed with the correct management.

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# SPONTANEOUS RUPTURE OF THE COMMON BILE DUCT (CBD) IN A 53-YEAR-OLD CAUCASIAN MALE ADMITTED WITH ACUTE PANCREATITIS

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**C**ommon bile duct (CBD) perforation has been associated with invasive procedures and blunt trauma. Spontaneous rupture, however, is an exceedingly rare occurrence in adults and has been reported less than 50 times in the world literature. This case report discusses a 53 year-old-gentleman who was admitted with acute pancreatitis secondary to gallstones. He continued to deteriorate clinically with bilious and enteric contents in an abdominal drain that was inserted to drain intra-abdominal collections. This resulted in this patient being taken to theatre for a diagnostic laparoscopy with a pre-operative diagnosis of gall bladder perforation. Intra-operatively, however, there was a 0.5 cm linear defect noted in the CBD. The gall bladder was intact. CBD was repaired using a T-tube drain followed by abdominal lavage. The patient recovered well and was discharged on day 49 post-operatively with T- tube in situ. Pre-operative diagnosis of CBD perforation can be challenging. It should be considered as a differential diagnosis in patients with bile present in the peritoneum. Early recognition is a key as prompt surgical intervention leads to a better prognosis.

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# CONGENITAL MID-GUT MAL-ROTATION WITH EPISODES OF PAIN FOR LONG PERIOD OF TIME

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**T**he prevalence of intestinal mal-rotation as a cause of abdominal pain is rare in adults and normally presents during infancy. Approximately 90% of patients with mal-rotation are diagnosed within the first year of life, 80% of them are diagnosed within the first month of life. The case was a 34-year-old pregnant woman whom was admitted to the hospital due to self-limited episodes of epigastric pain from young ages. The patient complained that the pains have worsened recently. Further clinical investigation leads us to invasive intervention due to signs of obstruction and the patient was transferred to operating room.

**Conclusion:** Mal-rotation of the mid-gut should be considered carefully in pregnant woman due to its similar signs and symptoms with pregnancy and it's necessary to be aware of the condition and management of it in order to decrease morbidity and mortality of the mother and neonate.

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# MEDICINE DISTRIBUTION IN SUDAN

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The strategy of price liberalisation and privatisation had been implemented in Sudan over the last decade, and has had a positive result on government deficit. The investment law approved recently has good statements and rules on the above strategy in particular to pharmacy regulations. Under the pressure of the new privatisation policy, the government introduced radical changes in the pharmacy regulations. To improve the effectiveness of the public pharmacy, resources should be switched towards areas of need, reducing inequalities and promoting better health conditions. Medicines are financed either through cost sharing or full private. The role of the private services is significant. A review of reform of financing medicines in Sudan is given in this study. Also, it highlights the current drug supply system in the public sector, which is currently responsibility of the Central Medical Supplies Public Corporation (CMS). In Sudan, the researchers did not identify any rigorous evaluations or quantitative studies about the impact of drug regulations on the quality of medicines and how to protect public health against counterfeit or low quality medicines, although it is practically possible. However, the regulations must be continually evaluated to ensure the public health is protected against by marketing high quality medicines rather than commercial interests, and the drug companies are held accountable for their conduct.

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# THE ANALYSIS OF POLYMORPHISM OF A1166C OF THE GENE OF AGTR AT PATIENTS WITH CHRONIC HEART FAILURE

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**Purpose:** To study distribution of genotypes of polymorphism of A1166C of a gene angiotensin's receptors of the 1<sup>st</sup> type (AGTR1) at patients of the chronic heart failure (CHF) with dysfunction of kidneys at persons of the Uzbek nationality.

**Research methods:** In total, 114 patients with the I-III functional class (FC) of CHF of the and the control group of 51 healthy faces of the Uzbek nationality were considered for carrying out the genetic analysis of polymorphism of A1166C of a gene of AGTR1 by standard polymerase chain reaction.

**Results of a Research:** The genetic analysis showed that distribution of genotypes corresponded HWD that testifies to a representativeness of selection and lack of various factors influencing genetic structure and also to the correctness of results of genotyping. Results of the carried-out analysis of distributions of genotypes of polymorphism of A1166C of a gene of AGTR1 at 114 patients CHF with the I-III FC in comparison with data of control group showed that this marker is independently not associated with development of CHF ( $p>0.05$ ). However at the same time, the reliable high frequency of occurrence of a genotype C/C in group of patients of CHF was noted ( $\chi^2=8.1$ ;  $p=0.04$ ) in comparison with population selection. The analysis of distributions of genotypes of polymorphism of A1166C of a gene of AGTR1 depending on the glomerular filtration rate (GFR) - more than 60 ml/min/1.73m<sup>2</sup> and less than 60 ml/min/1.73 m<sup>2</sup> showed that subgroups of patients CHF from GFR less than 60 and more than 60 ml/min/1.73 did not differ from control group ( $\chi^2<3.8$ ;  $p>0.05$ ). Besides, subgroups of patients also did not differ among themselves ( $\chi^2<3.8$ ;  $p>0.05$ ).

**Conclusions:** Polymorphism of A1166C of a gene of AGTR1 at persons of the Uzbek nationality is not associated with development of dysfunction of kidneys in patients with CHF.

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# A CASE OF TRAUMATIC ABDOMINAL WALL HERNIA SECONDARY TO SEATBELT INJURY: MULTIDISCIPLINARY MANAGEMENT BY DELAYED SURGICAL MESH REPAIR, COMPLICATED BY INTERVENING PREGNANCY

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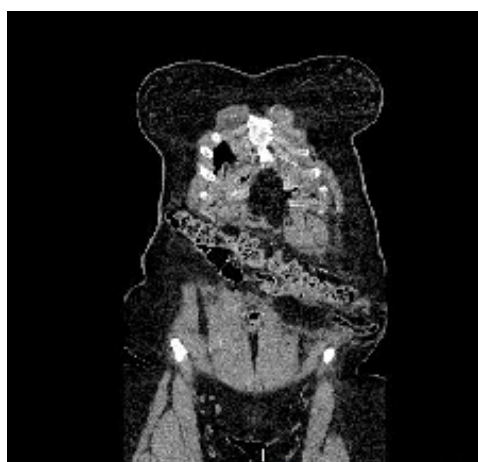
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**Background:** Traumatic abdominal wall hernia (TAWH) is a rare type of hernia occurring secondary to blunt trauma, with a reported incidence of 0.2% on presentation CT scan. There is currently no consensus regarding the optimum time for repair, though conservative management with serial imaging +/- expectant elective repair has been favoured.

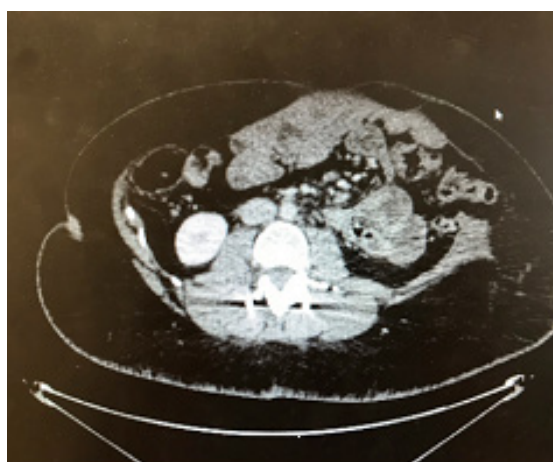
**Methods:** We present a case of a 20-year old female who was a seatbelt restrained rear-seat passenger in a road traffic collision and sustained two areas of Grade-5 TAWH along with bowel and colonic injuries. The patient was taken for an emergency laparotomy with initial conservative management of TAWH.

**Results:** Four years post-index admission, the patient's abdominal wall was closed using Strattice Mesh. Delay was due to the inability of the patient to reach the 100-110 kg goal operative weight and a pregnancy. During her pregnancy, the patient was managed by active surveillance involving the obstetric, plastics and trauma team.

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**Figure 1a:** Coronal CT slice demonstrating TAWH in the seatbelt distribution



**Figure 1b:** Axial CT slice demonstrating a Grade 5 TAWH

# A CASE REPORT ON RARE VARIANT OF ILNEB SYNDROME

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**C**ongenital interstitial lung disease with nephrotic syndrome and epidermolysis bullosa (*ILNEB*) is thought to be caused by mutation in the alpha 3 integrin gene (*ITGA3*). Alpha 3 integrin heterodimerises with beta 1 integrin and functions as a receptor for extracellular matrix proteins. It is important for cellular adhesion and is a component of fetal and adult tissues. To date, only six patients with *ILNEB* syndrome have been reported. All the patients carried homozygous *ITGA3* mutations and presented with severe phenotypes that culminated in death before two years of life. In this report, we present an atypical case of the *ILNEB* syndrome. A nine-year-old female child, second born of third degree consanguineous parents, came with complaints of insidious onset breathlessness for the past six months. Antenatal, natal and perinatal period were uneventful. The child was apparently normal till two years of age after which she developed blistering skin lesions that healed with scarring. She also had a history of passing foamy, frothy urine since the fourth year of life. On examination she had normal mentation, short stature, toe nail dystrophy, scarring at rest alopecia and epihora. She was tachypenic but could do her activities of daily living. Investigations revealed massive proteinuria suggestive of nephrotic syndrome, high resolution computed tomography suggestive of interstitial lung disease and skin biopsy revealed epidermolysis bullosa. The patient was given supportive treatment and her peripheral blood sample was subjected to next generation sequencing, which revealed a homozygous 3' splice site mutation in intron 13 of the *ITGA3* gene. Our sequencing efforts suggested the presence of an *ITGA3* pseudogene. This pseudogene could express a truncated but partially functional version of the *ITGA3* protein, which would explain the milder clinical symptoms in this patient. We are investigating this possibility. In summary, we describe a rare variant of the *ILNEB* syndrome associated with a homozygous mutation in the *ITGA3* gene. The presentation of milder phenotypes in this case is distinct from previously reported instances of this syndrome where all patients died by 19 months of age from multi-organ failure.

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# NECROTIZING ENTEROCOLITIS IN A NEWBORN PRETERM INFANT AND ROLE OF FEEDING: A CLINICAL CASE REPORT PRESENTATION

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**Statement of the Problem:** It's a clinical case presentation of a male preterm infant newborn, (+31 weeks) who was delivered in our hospital and transferred to our NICU because of prematurity, VLBW and need of respiratory support. Baby was detected with necrotizing enterocolitis (NEC) on 5<sup>th</sup> day of life, shortly after start of expressed milk feeding, which was detected early by the use of near infrared abdominal spectroscopy (NIRS). Baby was deteriorated clinically in a couple of hours and had intestinal perforation with peritonitis. So, abdominal exploration surgery with intestinal resection and end to end anastomosis was done urgently. Baby improved gradually and early feedings were started which were gradually increased up to full feedings with use of human fortified milk (HMF), probiotics and prebiotics.

**Findings:** The study stated the evidence-based feeding strategies guidelines for necrotizing enterocolitis (NEC) among very low birth weight infants and role of trophic feedings, probiotics, pre-biotics, micronutrients in prophylaxis, prevention and management of NEC.

**Recommendations:** Prematurity is the single greatest risk factor for NEC and avoidance of premature birth is the best way to prevent NEC. The role of feeding in the pathogenesis of NEC is uncertain, but it seems prudent to use breast milk (when available) and advance feedings slowly and cautiously. NEC is one of the leading causes of mortality and the most common reason for emergent GI surgery in newborns. NEC remains a major unsolved medical challenge, for which no specific therapy exists and its pathogenesis remains controversial. A better understanding of the pathophysiology will offer new and innovative therapeutic approaches and future studies should be focused on the roles of the epithelial barrier, innate immunity and microbiota in this disorder. Bioinformatics modeling is a new emerging strategy aimed at understanding the dynamics of various inflammatory markers and their application in early diagnosis and treatment.

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# HERNIA DEFECT CLOSURE WITH CORRESPONDING MESH SITE FIXATION ONLY IN LAPAROSCOPIC INGUINAL HERNIA REPAIR

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**Background:** The role of the mesh is very important in hernia repair especially in the endoscopic technique. The fixation by penetrating techniques is associated with a significantly increased risk of developing a post-hernioplasty pain syndrome. Tension free technique is associated with increased risk of mesh bulging or migration and recurrence. We have presented our technique in which the mesh is fixed by one suture at its centre and peripheral tension free.

**Methods:** This study included 75 male patients, suffering from inguinal hernias. Transabdominal preperitoneal (TAPP) technique was used in which the mesh was fixed by one suture at its centre and leaving peripheral mesh parts free. The intra and post-operative complications were recorded.

**Results:** The mean hospital stay was 1.2 days. The mean age was 41.3 years. The operative time ranged from 40 to 120 minutes. Mild bleeding was occurred in 9 patients (12%) during hernia sac dissection. Postoperative complications were mild inguinal pain in 5 patients (6.7%) for three weeks, and mild hydrocele in 10 (13.3%). No recurrence or mesh bulging or migration was noticed during the period of follow up (range 3 to 24 months).

**Conclusion:** Our procedure for mesh fixation during transabdominal preperitoneal laparoscopic inguinal hernia repair is easy, cost effective, associated with good results and free of complications.

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# THE STUDY OF ROLE OF MAGNESIUM SULPHATE VS. DIAZEPAM IN ECLAMPSIA

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Eclampsia is a life threatening condition with highest mortality and morbidity characterized by uncontrolled hypertension, unconsciousness fits albumin urea in the setting of preeclampsia.

**Study:** An extensive elaborative study carried out from 2005 to 2009, to study the therapeutic and prophylactic role of magnesium sulphate and Diazepam in Eclampsia.

**Aim:** To study the role of magnesium sulphate ( $MgSO_4$ ) vs. Diazepam in reducing maternal mortality ratio (MMR), neonatal mortality rate (NMR) in Eclampsia.

**Study Design:** This was a comparative study. Total 500 patients of fulminant pre-eclampsia /eclampsia including booked/unbooked patients attending Lady Willingdon Hospital were admitted, evaluated, assessed and put on  $MgSO_4$  vs. Diazepam divided in two groups. The results were compared which showed of  $MgSO_4$  has superior efficacy compared to Diazepam in improving overall mortality and morbidity in Eclampsia.

**Materials & Methods:** Total 500 patients of Eclampsia were admitted and studied in Unit-2 Lady Willingdon Teaching Hospital of King Edward Medical University Lahore managed in ICU according to a specially design proforma, protocol and were given  $MgSO_4$  and Diazepam for control of fits, Hydralazine, Labetalol and Isoket infusions for lowering blood pressure ( $MgSO_4$ ) A+B. Diazepam, with multidisciplinary involvement divided in two groups, compared and followed up.

**Results:** In Group A, there were 20 mothers and in Group B, 40 mothers died. It was observed that maternal mortality was significantly higher in Group B women as compared to that of Group A. i.e. (p-value=0.005).

**Conclusion:** The comparative study and of role of  $MgSO_4$  vs. Diazepam in reducing mortality, morbidity in maternities, neoneties, efficacy showed the superiority of magnesium sulphate as compared to Diazepam without any doubt.

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# AN EASY WAY TO ELIMINATE CAUSES OF COLLAGEN AND ALLERGIC DISEASES

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According to the traditional concept of the contemporary immunology, neither autoimmune diseases nor allergic diseases can be cured completely. Nevertheless, a fortunate coincidence led the author to discover a novel concept that eliminations of the causes of these diseases are possible. In other words, combinations of pathogenic antibodies with responsible cells, namely, cytolytic T lymphocytes in cases of autoimmune diseases and mast cells in cases of allergic diseases, can be decomposed by replacing the pathogenic antibodies with non-specific antibodies. In more detail, intradermal injections with a non-specific antigen preparation induce productions of non-specific antibodies in the body of the patient. Repetitions of the injections bring about an accumulation of them. Accumulated non-specific antibodies will occupy most of the receptors on the surface of responsible cells. When the accumulation reaches the sufficient level, virtually no pathogenic antibodies would remain on the receptors. That is, no causes of the diseases remain.

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# AWARENESS AMONG DOCTORS ABOUT THE ROLE OF SPEECH AND LANGUAGE PATHOLOGISTS IN THE MANAGEMENT OF DYSPHAGIA IN COPD PATIENTS

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**Objective:** To assess current awareness among doctors regarding the role of speech language pathologist (SLP) in management of dysphagia in chronic obstructive pulmonary disease (COPD) patients.

**Methodology:** Cross-sectional study involving 70 participants was conducted in two tertiary health care facilities in Islamabad and Rawalpindi, Pakistan. Information regarding their awareness was collected using self-administered questionnaire. SPSS was used to analyse the data.

**Results:** Out of 70 doctors, 28(40%) were not aware and 10(14.3%) disagreed while 32(45.7%) agreed that SLP plays an important role in the management of COPD patients.

**Conclusion:** There is good awareness about role of SLP in COPD management among the doctors and that these patients with dysphagia should be referred to SLP but less than half of them agree to do so, which is reflected in their practice. Consultants were more aware than those having lesser experience in their field.

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