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ABSTRACTS

Presentations of Sultan Qaboos University Clinico-Pathologic Conferences

Sultan Qaboos University College of Medicine & Health Sciences and Sultan Qaboos University Hospital, 2014–2015

عروض المؤتمرات الإكلينيكية الباثولوجية المنعقدة في جامعة السلطان قابوس

كلية الطب والعلوم الصحية في جامعة السلطان قابوس، ومستشفى جامعة السلطان قابوس، 2014-2015

Paroxysmal Nocturnal Haemoglobinurea

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Paroxysmal nocturnal haemoglobinuria (PNH) is a rare disorder characterised by deficiency of glycosylphosphatidylinositol-anchored complement regulatory proteins. Manifestations range from indolent to life-threatening, including Coombs'-negative intravascular haemolysis, abdominal pain associated with smooth muscle dystonia, renal impairment, cytopaenia and thrombosis (typically affecting the portal and mesenteric veins and occasionally the arterial veins). Testing should be done in cases with such manifestations and in those with bone marrow failure or cytopaenia of unknown aetiology. Flow cytometry is the gold-standard test for this condition. Management includes observation for asymptomatic cases, transfusion, anticoagulation, eculizumab administration and bone marrow transplantation (BMT). PNH is rare in Oman and only six cases have been diagnosed at the Sultan Qaboos University Hospital in Muscat, Oman. Of these cases, four underwent BMT, while two were prescribed eculizumab. We present a complicated case of flow cytometry-diagnosed PNH presenting with thrombosis during pregnancy and progressing to renal impairment and bone marrow failure.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 13 November 2014 with the title "Go with the flow".

Percutaneous Transvenous Mitral Commissurotomy

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Percutaneous transvenous mitral commissurotomy (PTMC) is the preferred alternative to surgical commissurotomy among suitable patients with severe rheumatic mitral *stenosis*. PTMC is a minimally invasive procedure to correct an uncomplicated mitral *stenosis* by dilating the valve using a balloon. Pregnancy in women with mitral *stenosis* is associated with a marked increase in maternal morbidity and adverse fetal outcomes. A multidisciplinary approach reduces mortality and morbidity during the peripartum stage of pregnancy. If symptoms persist despite optimal medical treatment, PTMC should be considered. We report a case of a woman who presented at the Sultan Qaboos University Hospital (SQUH) in Muscat, Oman, with symptomatic severe mitral *stenosis* during pregnancy. She was managed medically after refusing intervention. However, after delivery she underwent a successful PTMC as the first case in SQUH. One month after the procedure, the patient showed dramatic improvement, both clinically and on echocardiography.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 20 November 2014 with the title "Risky business".

Developmental Consequences of 22q11.2 Microdeletion Syndrome

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Raising awareness of 22q11.2 deletion syndrome amongst physicians is crucial. The clinical phenotype of this syndrome is due to haploinsufficiency of multiple dosage-sensitive genes, resulting in the disruption of numerous embryonic developmental processes. These include cardiovascular/palatal anomalies, immunodeficiencies, hypocalcaemia and cognitive/psychiatric disorders. Although 22q11.2 deletion syndrome is the most common microdeletion condition in humans, it remains the most underdiagnosed because of its remarkable variability and expression among affected individuals. We present a case series of six children with 22q11.2 deletion syndrome seen at the Genetics Clinic of the Sultan Qaboos University Hospital in Muscat, Oman. These patients had molecularly-confirmed 22q11.2 deletion which was diagnosed by either fluorescence *in situ* hybridisation or array-comparative genomic hybridisation in three cases each. This series highlights a holistic approach to the diagnosis and management of 22q11.2 deletion, including the use of a multidisciplinary team. An early diagnosis also provides the best opportunity to optimise patient care.

This case series was presented at the Sultan Qaboos University Clinico-Pathological Conference on 11 December 2014 with the title "Fishing for a good catch".

Development of Obstructive Sleep Apnoea in Patients with Pierre Robin Sequence

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Pierre Robin sequence (PRS) is a condition characterised by the triad of micrognathia/retrognathia, *glossoptosis* and a 'V'-shaped palate with or without a cleft palate. It occurs in approximately one in 8,500 births. Infants in the immediate and early postnatal period may experience varying degrees of upper airway obstruction leading to the development of obstructive sleep apnoea (OSA). Mandibular distraction osteogenesis (MDO) has a high success rate and is now the gold standard in the management of moderate to severe OSA in PRS patients. We present a case of PRS with severe OSA that was managed surgically with MDO and resulted in the immediate successful elimination of the OSA. This case highlights the clinical appearance and pathogenesis of the condition, as well as the importance of preoperative full airway and anaesthetic assessments. In addition, PRS cases that were managed with MDO in Oman between 2008–2014 are also reviewed.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 25 December 2014 with the title "Whoever saves a life, it is as if he saved the life of all mankind".

Lung Transplantation: *First patient from Oman since the early 1990s*

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We report a 19-year-old Omani male who was the first patient from Oman to undergo a lung transplant since the early 1990s. He was born to consanguineous parents and was diagnosed with bilateral bronchiectasis at three years old. His mother and sister had been diagnosed with the same condition, leading to his mother's early death. Computed tomography performed at the Sultan Qaboos University Hospital in Muscat, Oman, showed extensive cystic bronchiectasis as well as areas of air trapping and partial volume loss. In March 2013, he underwent bilateral lung transplantation at the King Faisal Specialist Hospital in Riyadh, Saudi Arabia. His postoperative course was protracted with episodes of acute rejection. To date, he has recovered well, despite several respiratory infections. The unique features of this transplant included short cold preservation time (4–8 hours), continuous external exposure of the organ, complex surgery (six anastomoses) and damaged donor lungs. Lung transplantation challenges include costs/logistics, cultural/religious aspects, donor shortages, long waiting lists and the need for frequent follow-up.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 8 January 2015 with the title "A new breath, a new life".

Intraoperative Pulmonary Embolism

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An intraoperative pulmonary embolism (IPE) can be catastrophic. Risk factors include major surgery as well as pelvic and lower extremity trauma. Thromboprophylaxis is recommended and inferior *vena cava* filters are reserved for patients with contraindications to prophylaxis. Whenever possible, neuraxial anaesthesia should be considered. Specific electrocardiogram changes are seen in approximately 25% of affected patients. Echocardiography findings of a massive IPE include acute right ventricular dilatation and dysfunction with leftward septal displacement. The initial treatment comprises cardiorespiratory support. Definitive treatment with anticoagulants and thrombolysis carry the risk of bleeding while conservative approaches can result in life-threatening obstructive shock. Surgical embolectomies and direct catheter-based approaches are rare. Early extracorporeal membrane oxygenation (ECMO) should be considered in order to reduce pulmonary vascular resistance and improve haemodynamics and tissue oxygenation. However, ECMO requires systemic anticoagulation as well. Prevention and a high index of suspicion in high-risk patients cannot be overemphasised.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 15 January 2015 with the title "Walking on the tightrope".

Syndromic Hypoparathyroidism with Septic Shock

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A six-week-old male baby was transferred from a peripheral hospital to the Sultan Qaboos University Hospital (SQUH) in Muscat, Oman, after having hypocalcaemic seizures. The baby was born to consanguineous parents at 35 gestational weeks with intrauterine growth retardation. A physical examination revealed dysmorphic features suggestive of Sanjad-Sakati syndrome. He was subsequently found to have severe hypocalcaemia and hyperphosphataemia and was diagnosed with hypoparathyroidism. His condition was managed by intravenous calcium infusion but this was interrupted due to difficulties with intravenous access. His treatment was further complicated by right femoral line displacement into the abdominal cavity, requiring an urgent laparotomy. The child then developed septic shock resulting from *Klebsiella* and *Candida albicans* infections. He was prescribed broad-spectrum antibiotics and antifungal medication and was successfully extubated after five weeks. He developed cholestasis, which gradually improved, and underwent an operation for bilateral cataracts before being discharged.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 22 January 2015 with the title "Outwit, outplay and outlast".

Phosphaturic Mesenchymal Tumour Treated with Octreotide and Cured by Surgical Removal

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Tumour-induced osteomalacia (TIO) is a rare disorder associated with the development of osteomalacia as a result of renal phosphate wasting. The latter results from increased circulating levels of fibroblast growth factor 23 which is secreted by a variety of different mesenchymal tumours that may be very small and difficult to find. This association was first reported by McCance in 1947. We report a case of tumour-induced hypophosphataemia and severe osteomalacia presenting to the Sultan Qaboos University Hospital, Muscat, Oman, whereby the tumour was localised with the use of positron emission tomography and octreotide scintigraphy. The patient responded clinically and biochemically to a short trial of octreotide and was cured by the surgical removal of the tumour.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 5 February 2015 with the title "Seen, but again, not observed".

Commercial Kidney Transplant Complicated by Severe Infection

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A seven-year-old boy received a commercial renal transplant due to chronic renal failure secondary to congenital posterior urethral valves and bilateral hydronephrosis. The immediate post-transplant period was uneventful. However, over the course of the following six months he had several episodes of diarrhoea associated with rising creatinine levels. His parents refused a renal biopsy and the child was managed conservatively with dosage modifications to his immunosuppression medication and antibiotic treatment. Six months after the transplant, his condition deteriorated. He developed persistent fever, respiratory distress and a reduced urine output necessitating admission to the intensive care unit. An ultrasound showed an oedematous graft kidney with multiple hypoechoic regions while a computer tomography angiogram confirmed multiple renal infarcts. He underwent a life-saving graft nephrectomy which showed widespread parenchymal and angioinvasive mucormycosis with infarction. Following a six-month postoperative period that included several surgical debridements for mucormycosis, the child was alive and undergoing dialysis again.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 19 February 2015 with the title "For sale".

Traumatic Brain Injury Leading to Cognitive and Behavioural Impairment in a 69-Year-Old Man

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The geriatric age group is rapidly growing in Oman and cognitive and behavioural challenges should be recognised among the various disorders affecting these individuals. We report a 69-year-old male who was referred to the Memory Clinic at Sultan Qaboos University Hospital in Muscat, Oman. The patient was suffering from behavioural changes of one month's duration following recovery from a coma after a road traffic accident. He became increasingly irritable, argumentative and forgetful. He was easily provoked, had threatened to kill himself and reported visual and auditory hallucinations as well as visuospatial disorientation. He could no longer recognise close family members and experienced functional impairment in daily activities. This case highlights the neuropsychiatric manifestations of traumatic brain injuries. In addition, it emphasises the importance of comprehensive assessment to differentiate between the sequelae of traumatic brain injuries and a neurodegenerative disorder like Alzheimer's disease, which can also be triggered by head trauma.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 5 March 2015 with the title "A walk down memory lane".

Overview of Cystic Fibrosis in Sultan Qaboos University Hospital with Focus on Ivacaftor Therapy

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We present four patients with cystic fibrosis (CF) with full manifestations of the disease and different outcomes modified by available therapies. All four patients were diagnosed with CF in infancy, presenting at the Sultan Qaboos University Hospital in Muscat, Oman, with failure to thrive and chronic respiratory symptoms. Their sweat chloride levels were above 60 mmol/L and they were prescribed enzyme replacement therapy (pancrelipase), high-caloric diet supplement vitamins and daily chest physiotherapy. Subsequently, the patients developed persistent *Pseudomonas* lung infections with bronchiectasis. They were administered regular nebulised tobramycin, 7% saline and dornase alfa but deteriorated clinically. Two of the patients passed away due to irreversible respiratory failure and severe lung infections at eight and 14 years old, respectively. The remaining two patients were given ivacaftor which improved their lung function by 20% and their weight by an average of 5 kg. They subsequently experienced an overall improvement in quality of life.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 12 March 2015 with the title "A new dawn for cystic fibrosis patients".

Blood Cultures

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Blood cultures are essential diagnostic tools and blood samples are arguably the most critical samples in the microbiology laboratory. Once a blood culture bottle reaches the laboratory, it is processed through a series of tests involving different types of equipment. This process takes a minimum of three days. Sometimes, a blood culture grows organisms that are not really present in the patient's blood. This is known as blood culture contamination, which can adversely affect patient management, prolong hospital stay and incur unnecessary costs. This presentation describes how blood cultures are processed in the microbiology laboratory. Three different clinical cases are presented to highlight key learning points. In particular, blood culture contamination is discussed, emphasising the adverse effects it causes and how it can be prevented.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 2 April 2015 with the title "Blood cultures: What happens in the dark tower?".

Acquired Haemophilia A

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Acquired haemophilia A is characterised by the presence of autoantibodies against endogenous factor VIII and presents with spontaneous bleeding *diathesis* associated with an isolated prolonged activated partial thrombosis time (aPTT). We report a 54-year-old man who presented at the Sultan Qaboos University Hospital in Muscat, Oman, with a history of progressive left elbow swelling associated with skin discolouration and limited movement. Investigations revealed an isolated prolonged aPTT of 154 seconds, which did not correct during mixing studies; very low factor VIII levels (<0.01 IU/mL); and high Bethesda anti-factor VIII antibody titres of 33.92 Bethesda units. The patient was given factor VIII:C concentrates along with recombinant factor VIIIc daily for a week to control the underlying bleeding *diathesis*. Eradication of factor VIII inhibitor was achieved with combined prednisolone/cyclophosphamide over one month along with four weekly 375 mg/m^2 doses of rituximab. This resulted in aPTT and factor VIII:C normalisation without the inhibitor.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 9 April 2015 with the title "The enemy within".

Awareness of Early Symptoms of Colorectal Cancer

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Colorectal cancer (CRC) is the third most commonly diagnosed cancer in males and the second in females worldwide. The survival rate from CRC depends on the stage at which the diagnosis is made; in many developing countries, there is often a delay in diagnosis. The delay can occur due to patients, doctors and/or the healthcare system. Primary care is the first point of contact for patients with CRC, however early-stage patients may delay presenting or present with non-specific symptoms. This presentation highlights the importance for doctors in primary care facilities to be alert for suspicious symptoms of CRC and investigate or refer cases as early as possible. Doctors should also follow national or international CRC guidelines to manage such patients. Furthermore, it is crucial that a national healthcare strategy be developed to increase public awareness about CRC risk factors and early symptoms.

This case was presented at the Sultan Qaboos University Clinico-Pathological Conference on 16 April 2015 with the title "Be alert before it is too late".