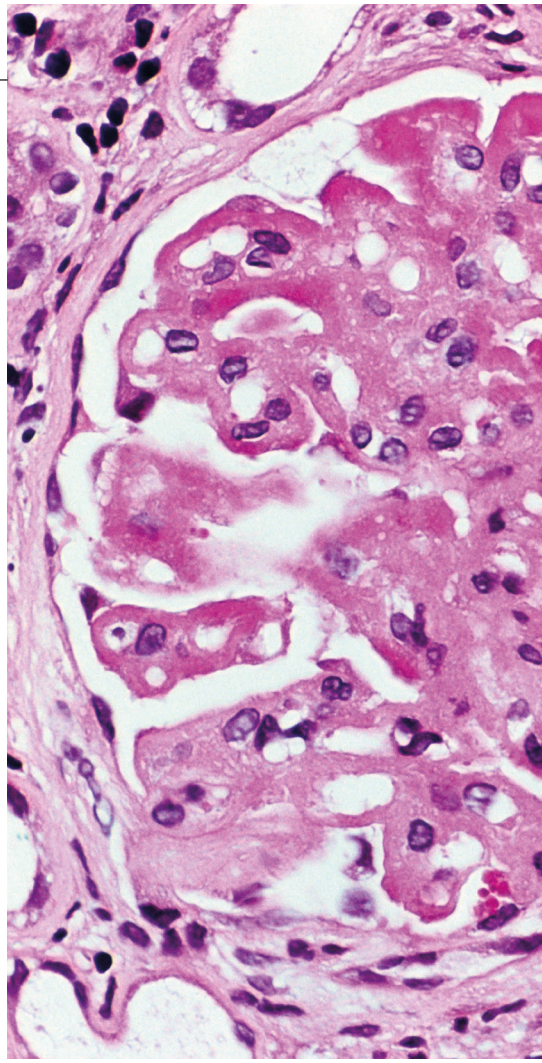


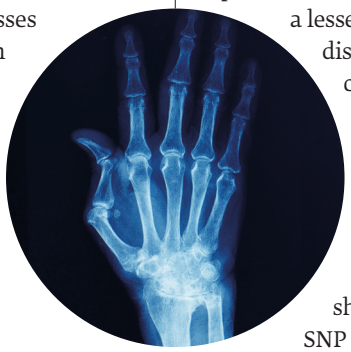
BRITISH JOURNAL OF BIOMEDICAL SCIENCE

ISSUE 1 2020 – SYNOPSIS

Deputy Editor **Tony Rhodes** provides a brief glimpse of the articles on offer in the first issue of 2020.

Firstly, in his editorial to this issue Editor Andrew Blann provides a review of what we have learnt from the articles published in the journal in 2019. The editorial is useful reading for those wishing to publish, as it captures the essence of what the journal is about and provides an excellent overview of the type of article and subject material published so far. No doubt it will also be a valuable point of reference for those of us undertaking journal-based learning as part of our continuing professional development.

Single Nucleotide Polymorphisms (SNPs) and/or microRNA (miRNA) and their pathological associations feature in many of the articles published in this issue of the journal (Gupta et al, Bahreini et al, Ramezani et al, Tian et al, Adbulla et al), whilst two of the articles involve autoimmune disease processes in glomerulonephritis (Tian et al) and rheumatoid arthritis (RA) (Huang et al). In addition, a total of four of the studies published in this issue focus on epithelial cancers (Gupta et al, Bahreini et al, Ramezani et al, El-Bendary et al).



SNP, miRNA and cancer

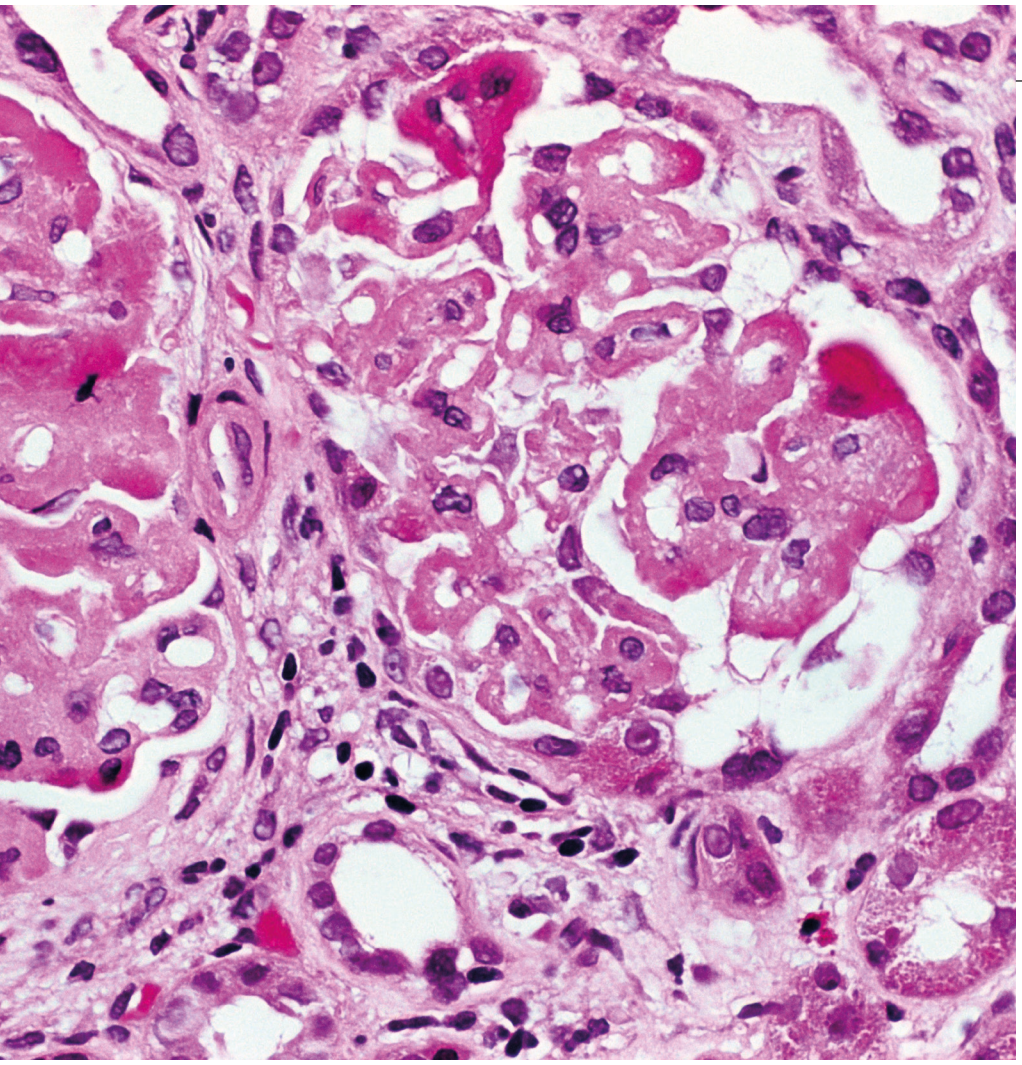
Gupta et al show that polymorphisms in the XRCC4 gene responsible for DNA repair are associated with cervical cancer, Bahreini et al show how a polymorphism in the mi-RNA-559, rs5840758, is linked to breast cancer, whilst Ramezani et al investigate how circulating miRNA can be used as a blood-based marker of breast cancer, showing the differential expression of the circulating miRNA, with -miR-125a-3P decreasing and -miR–125b levels increasing in the disease, respectively.

Autoimmune disease processes in glomerulonephritis and rheumatoid arthritis

Membranous nephropathy (MN), the underlying disease process responsible for the nephrotic syndrome in adults, is predominantly idiopathic (IMN), and to a lesser degree secondary to other diseases (SMN). IMN is now considered by many to be an autoimmune disease, in which autoantibodies target the M-type phospholipase A2 receptor (PLA2R) on glomerular podocytes. Recent studies show an association between SNP of PLA2R1, and in this issue of the journal Tian et al investigate the expression of five of these SNP in Chinese patients with IMN and SMN.

RA is a chronic and systemic autoimmune disease in which there is destruction of synovial tissue of the joints due to a chronic inflammatory process, resulting in loss of joint function. Whilst the exact pathogenesis of RA has yet to be established, cytokines and inflammatory mediators play a key role. The 14-3-3 family of proteins, which have a range of functions, to include involvement in inflammation, are known to be important in the development of RA, with serum 14-3-3η upregulating cytokines and allowing systemic inflammation to persist. Recently the protein has become used clinically as a marker for the disease in addition to existing markers, such as anticitrullinated peptide antibodies (ACPAs) and the rheumatoid factor (RF).

In addition, the pro-inflammatory cytokine, high mobility group box-1 (HMGB1) plays a role in the pathogenesis of many diseases, including RA. In this



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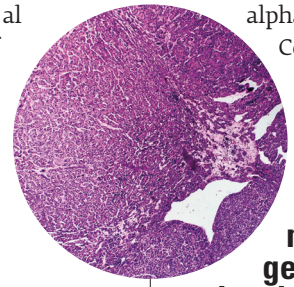
issue of the journal, Huang et al set out to determine which of these markers alone or in combination provide the best data for the diagnosis of RA.



Methylation of tumour suppressor genes and E-cadherin in hepatocellular carcinoma

Cancer of the liver (hepatocellular carcinoma) is a leading cause of death in many parts of the world and is frequently associated with long-standing infection with either the hepatitis B virus (HBV) or hepatitis C virus (HCV). In this issue of the *British Journal of Biomedical Science*, El-Bendary et al, investigate the methylation of tumour suppressor genes (RUNX3, RASSF1A) and the cell adhesion molecule, E-cadherin, in HCV related liver cirrhosis and hepatocellular carcinoma. They report the methylation status of the E-cadherin gene to be an independent risk factor for cases of HCV-associated hepatocellular carcinomas with low alpha-fetoprotein (AFP). Consequently, this finding may be of diagnostic value in such cases.

SNP for collagen and metalloproteinase genes and their involvement in keratoconus

Keratoconus, a corneal disease typified by thinning of the cornea and resulting in irregular astigmatism and decreased vision, may be linked to a decreased amount of total collagen matrix. Matrix metalloproteinase-9 (MMP9) and gelatinase are the main matrix-degrading enzymes produced by the corneal epithelium. The balance of MMP9 activity is in turn regulated by tissue inhibitors of matrix metalloproteinase (TIMPs). In their article, Abdullah et al hypothesise that SNP for collagen IV, MMP9 and TIMP-1 genes play a role in the pathogenesis that leads to the corneal thinning seen in keratoconus.

"If further studies can verify these results, it will increase our understanding of the disease"

Alkaline phosphatase levels and epilepsy

Rawat et al, in their biomedical science in brief, show that the raised alkaline phosphatase (AP) levels frequently seen in patients with epilepsy are related to the frequency of recurring epileptic seizures and how recently they occurred and not due to the metabolic and pharmacological interactions of epileptic drugs, as previously thought. They were able to show this by comparing results in a previously untreated, drug-naive, group of patients compared to a group already receiving medication. If further studies can further verify these results and elucidate the mechanisms involved, it will clearly increase our understanding of the disease.

Case study of Madelung disease

Hoxha et al from Tirane, Albania, present a case of Madelung disease; an extremely rare condition with an incidence of just 1/250,000 and occurring more commonly in the Mediterranean populations. It is characterised by symmetrical abnormal growth of adipose tissue, occurring particularly around the neck, upper limbs and chest. The main differential diagnosis is Cushing's syndrome, which was ruled out following laboratory tests for cortisol levels. In this case report, the authors summarise what is known about the disease and the investigations taken to arrive at the definitive diagnosis.

CPD

Any of the above may be the subject of Journal-based learning. BMS