

**Background and aims** To observe the effects of long term hydroxychloroquine treatment on blood lipid and left ventricular function of systemic lupus erythematosus(SLE) patients.

**Methods** 72 patients with SLE were randomly divided into two groups: Hydroxychloroquine treatment group(n=36)and non-hydroxychloroquine group(n=36). The level of blood lipid, left ventricular end-diastolic diameter (LVEDD), left ventricular end-systolic diameter (LVESD), interventricular septum thickness(IVST), *left ventricular* posterior wall thickness (LVPWT), fractional shortening rate(FS), left ventricular ejection fraction(LVEF), E/A were measured before, 6 month, 12 month and 2 years after the treatment.

**Results** The long term applies of hydroxychloroquine can bring statistically different of TC, TG, LDL and HDL to SLE patients. LVEDD, LVWPT and E/A were statistically different ( $p<0.05$ ) before and after hydroxychloroquine were used.

**Conclusions** The long term applies of hydroxychloroquine can improve the lipidic metabolism and left ventricular function in SLE patients.

## Genetics, epigenetics, omics, biomarkers and personalised medicine in SLE and autoimmunity

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### UTILITY OF SERUM FERRITIN AS A MARKER OF DISEASE ACTIVITY IN CHILDHOOD SYSTEMIC LUPUS ERYTHEMATOSUS

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10.1136/lupus-2017-000215.262

**Background and aims** To assess the usefulness of serum ferritin levels as a marker of disease activity and organ involvement in childhood systemic lupus erythematosus (cSLE) and to screen children with SLE for subclinical macrophage activation syndrome.

**Methods** Consecutive children who met the criteria of SLICC were enrolled. All patients interviewed and assessed for disease activity using SLE disease activity index (SLEDAI). Biochemical and serological tests including serum ferritin level and markers of disease activity and macrophage activation syndrome (MAS) including LDH, AST, triglyceride and CD25 were measured by standard laboratory procedure.

**Results** A total of 29 (24 female) SLE patients with a mean age of 10.9 ( $\pm 2.9$ ) years and mean of disease duration of 4 ( $\pm 2.4$ ) years were included. The most frequent manifestations were musculoskeletal in 25 patients followed by haematological in 15 then renal involvement in 13 patients. Twenty patients had active disease (SLEDAI  $>4$ ). Serum ferritin level was correlated significantly with SLEDAI ( $p<0.0001$ ) and markers of MAS (LDH, AST, triglyceride and CD25) and negative correlation with fibrinogen ( $p 0.02$ ). Interestingly, serum ferritin was weakly correlated with ESR but no correlation with CRP and proteinuria. Two patients confirmed to have MAS.

**Conclusions** Serum ferritin is a simple and probably a good marker of disease activity and screening for MAS in cSLE.

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### UPDATE ON SLE-HLA ASSOCIATION: CONTRIBUTION OF ALLELE-SPECIFIC EXPRESSION IN ADDITION TO AMINO ACID CHANGES IN HLA ALLELES

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10.1136/lupus-2017-000215.263

**Background and aims** A genetic contribution of the human leukocyte antigen (HLA) genes to SLE has been well documented (e.g., OR=1.65, Heritability explained by HLA=2%; *Nat Genet* 2016). To understand the association in HLA loci within the major histocompatibility complex (MHC) region in large cohorts genotyped for MHC SNPs, we developed an ethnicity-matched HLA reference panel (*PLoS One* 2014).

**Methods** Using the HLA imputation and various statistical approaches, we investigated HLA amino acid residues, HLA classical alleles and MHC SNPs simultaneously, and identified that the changes in amino acid positions 11, 13 and 26 of HLA-DRB1 explained the entire HLA association (*Nat Commun* 2014; *Nat Genet* 2016). Additionally, all the protein-coding HLA-DR beta genes (*HLA-DRB1*, *HLA-DRB3*, *HLA-DRB4* and *HLA-DRB5*) with similar functions were further investigated using imputation-based conditional regression and haplotype analyses. *HLA-DRB1* was solely associated with SLE and accounted for the associations of the other HLA-DR beta genes (*PLoS One* 2016). Finally, we measured allele-specific expression of *HLA-DRB1* in blood cells by RNA sequencing followed by an allele-specific read mapping method.

**Results** Strong allele-specific expression among *HLA-DRB1* classical alleles was observed, which caused relatively unequal expression of two heterozygous alleles in individuals. Disease association models, fitted by logistic regression including either the copy number or both the copy number and the relative expression of each allele as predictors, revealed that the SLE association was significantly better explained by adding the variables for the relative expression.

**Conclusions** These findings indicate that both the qualitative and quantitative effects of *HLA-DRB1* variants are driving SLE (Figure).

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### PEARL: PATHWAY EXPLORATION AND ANALYSIS IN RENAL DISEASE IN THE ACCELERATING MEDICINE PARTNERSHIP (AMP) LUPUS NETWORK

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10.1136/lupus-2017-000215.264

**Background and aims** Despite treatments, a substantial proportion of lupus nephritis (LN) patients progress to end stage renal disease and death. Detailed transcriptomic analyses of LN kidneys may identify new therapeutic targets. Our goal is to demonstrate the feasibility of single cell and low-input transcriptomic analyses of LN kidney and urine cells.

**Methods** Cells from urine and renal biopsies performed for clinical diagnosis from inform-consented patients (1 class III, 3 class IV+V, 1 class V) and 1 control (healthy part of tumour nephrectomy) were isolated, frozen, sorted and analysed by RNAseq.