DEEP PHENOTYPING OF PSORIASIS BASED ON LIPID METABOLISM RELATED GENES BY INTEGRATIVE SYSTEMS ANALYSIS

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Methods: We performed molecular subtyping and association analysis of psoriasis from Gene Expression Omnibus (GEO). The training sets included GSE13355 cohort (N = 58), GSE14905 (N = 26) and GSE30999 cohort (N = 81), while the GSE54456 (N = 92) was selected as the validation set. Energy metabolism-related genes were clustered into GSVA index. Based on these psoriasis lipid metabolism-related genes, we conducted consensus molecular subtyping with nonnegative matrix factorization (NMF). Finally, Gen set variation analysis (GSVA) was used to score individual subtypes against the metabolism-related gene sets, and each group got a GSVA index.

Results: Total 104 lipid metabolism-related genes were clustered into three subgroups (Figure 1A). GSVA revealed the lipid metabolism with the correlation in subtypes. There was no statistical difference was identified between the subtype 1 and subtype 2 (P > 0.05, Figure 1B), so we merged the two types as a new Sub1, another subtype was named as the new Sub2 (Figure 1C). We finally identified 2 distinct subtypes of psoriasis in the two types (P > 0.05, Figure 1B), so we merged the two types as a new Sub1, another subtype was named as the new Sub2 (Figure 1C).

Conclusion: This study established a new classification system that was based on the gene expression profile of lipid metabolism related genes.

REFERENCES:

DETECTION OF ABCG2 VARIANTS IN ENCODING OF URATE TRANSPORTERS ASSOCIATED WITH THE HYPERURICEMIA IN HAEMODIALYSIS PATIENTS

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Methods: In this retrospective one centre study 167 patients (age=79.8±10.3, female=74) with end-stage CKD (pre-dialysis n=86, dialysis n=79) were collected. Peak urate levels were 456.3±113.6 μmol/l in pre-dialysis and 572.3±114.6 μmol/l in dialysis. ABCG2 coding regions were analyzed from genomic DNA, as we described previously (1). The reference sequence was defined as version ENST00000237612.7, the reference protein sequence was defined as Q9UNQ0. The chi-square goodness-of-fit test was used to compare minor allele frequencies (MAF), and the log-rank test was used to compare empirical distribution functions.

Results: In the CKD cohort, 15 intronic and seven non-synonymous allelic exonic variants were detected: two common (rs2231137/p.V12M; rs2231142/p. Q141K), five ultra rare and/or rare (rs142634180/p.R45Q, rs759726272/p.M131I, rs140207606/p.R236X, rs138606116/p.G354R, rs138892154/p.A607V), and one novel (p.E344D). Common variant p.V12M, previously reported as protective