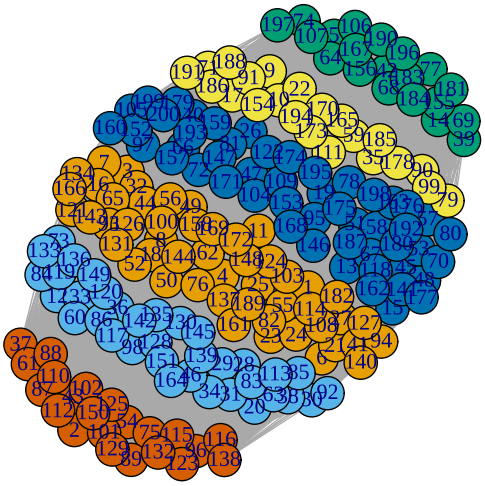
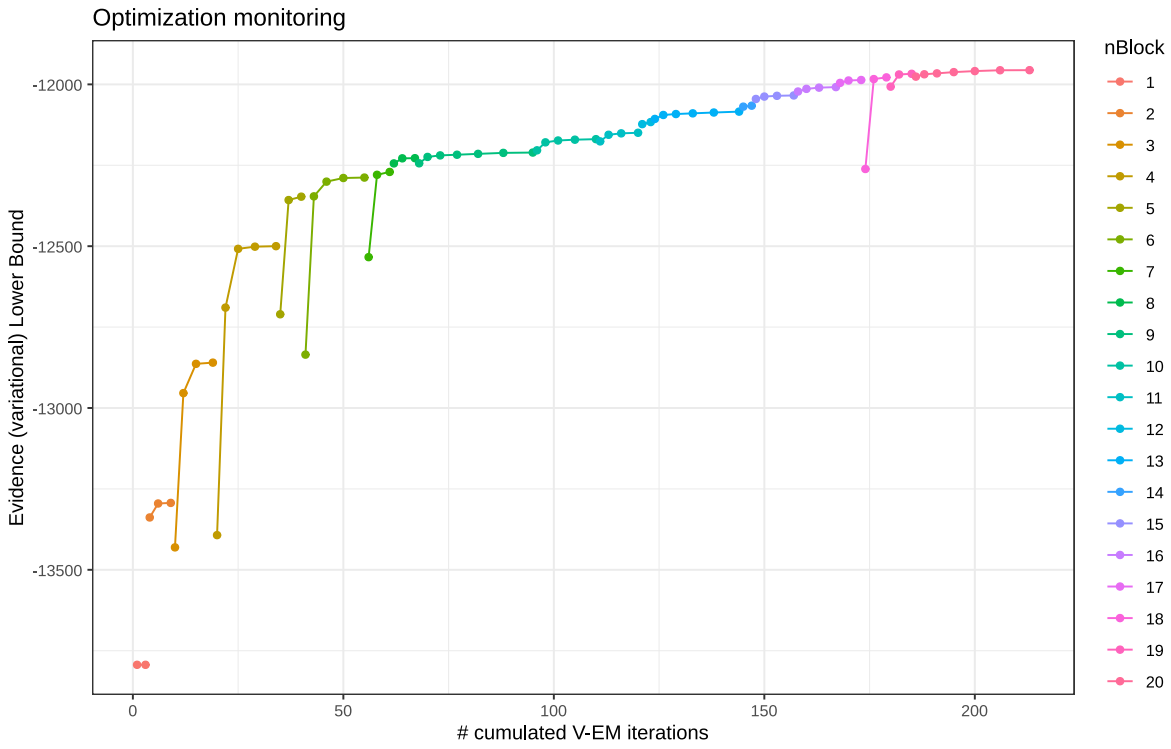
结果分析

训练结果

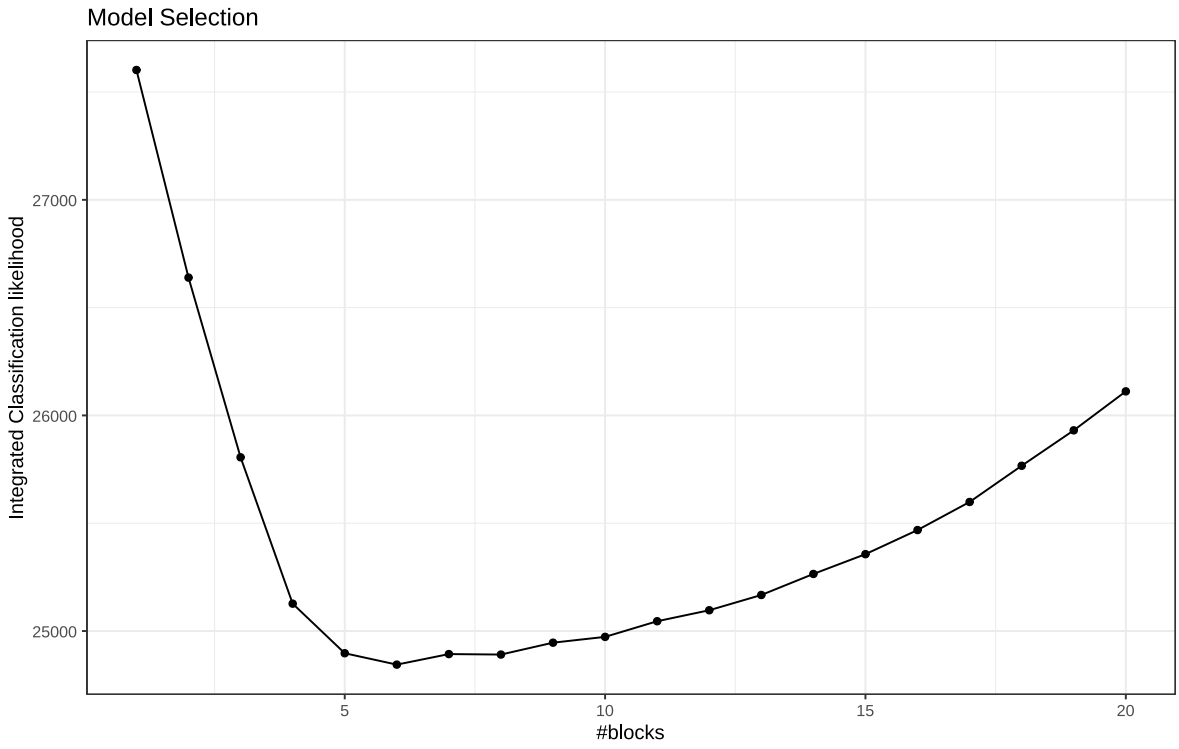


其中

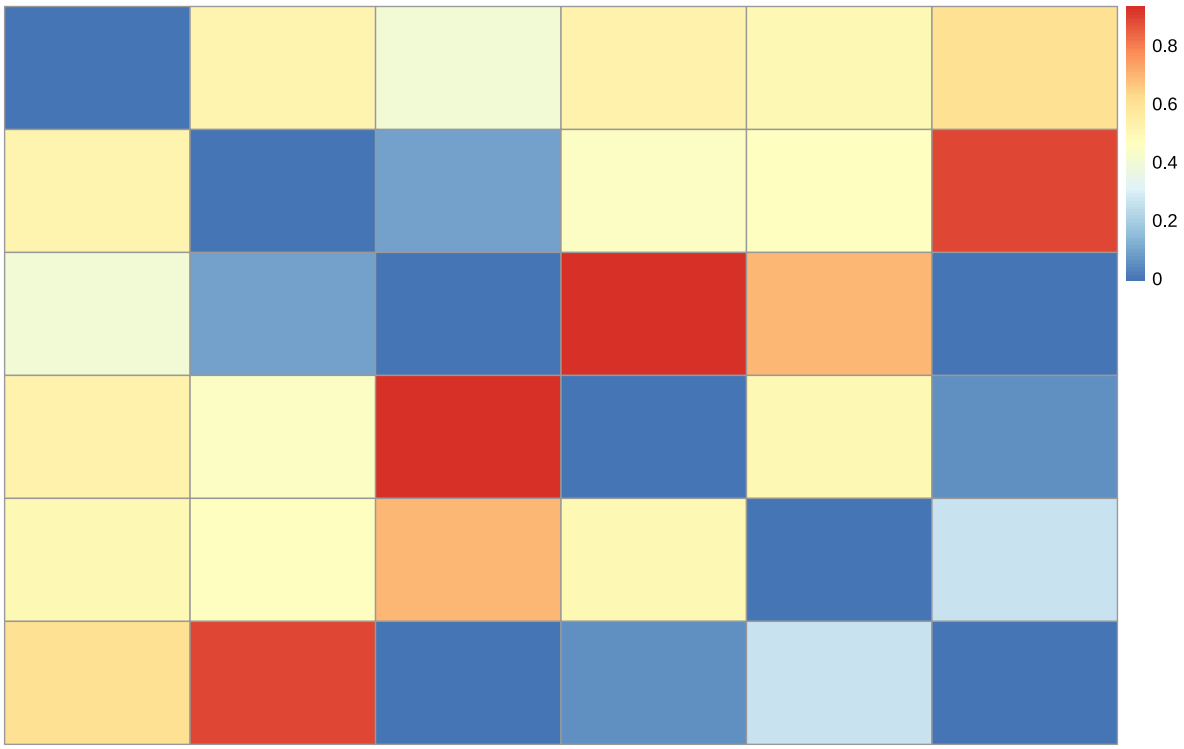
- 深红色为簇6
- 浅蓝色为簇2
- 棕色为簇1
- 深蓝色为簇5
- 黄色为簇4
- 绿色为簇3



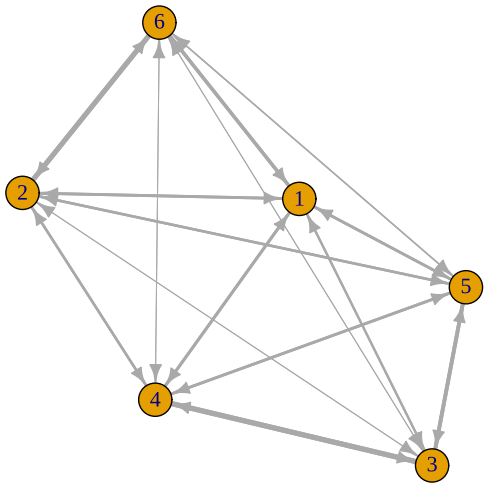
对于给定不同簇数，EM迭代算法给出ELBO迭代图：越高越好，可看出ELBO整体上呈现上升趋势



但是如果考虑簇内关系的话，簇数为6时vem算法给出最小值

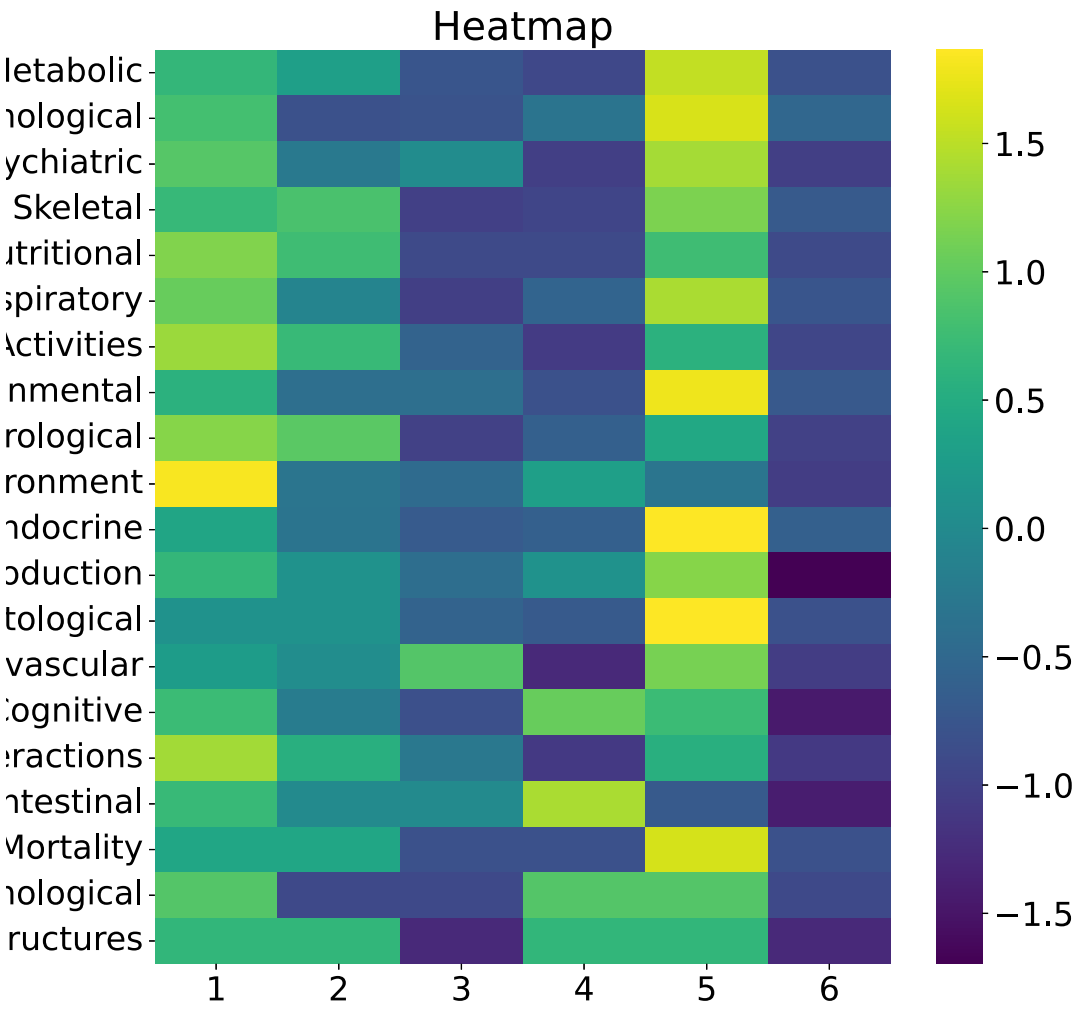


簇与簇之间的关联如下

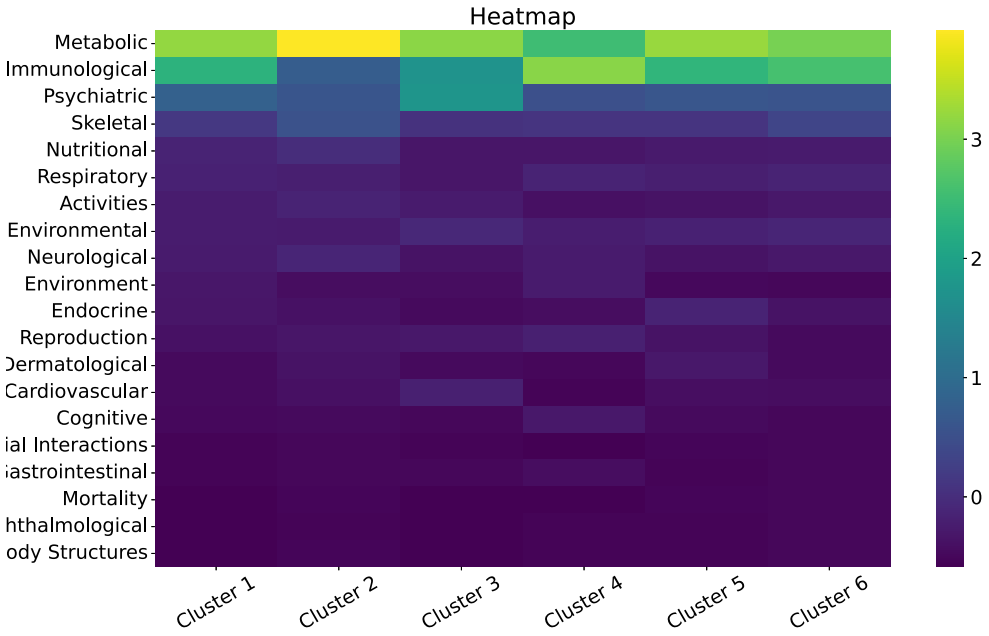


结合表型分析

对于每个簇，我们分析簇内SNP相关的表型性状；给出如下热图

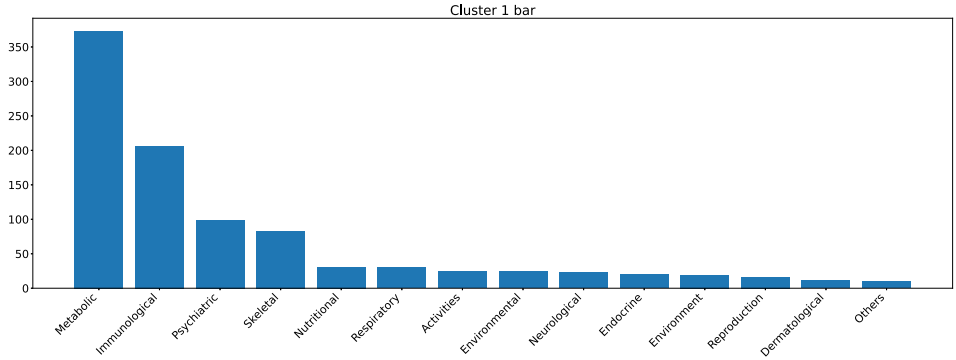


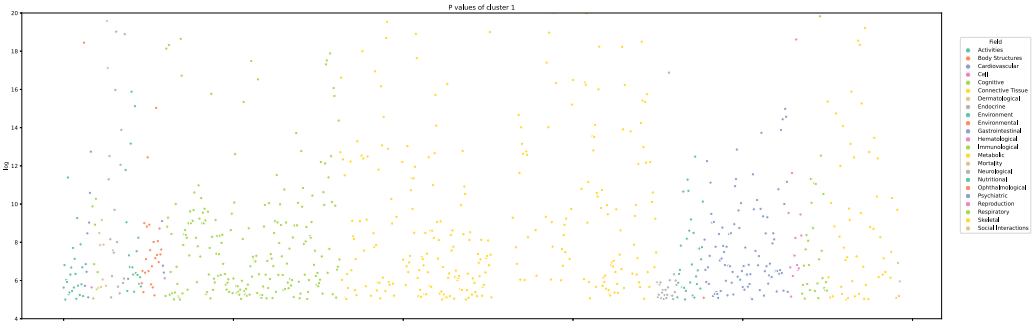
该图通过表型标准化展现了簇与簇之间在不同性状之间的差异，可发现簇1，5具有较强的信号，值得继续分析

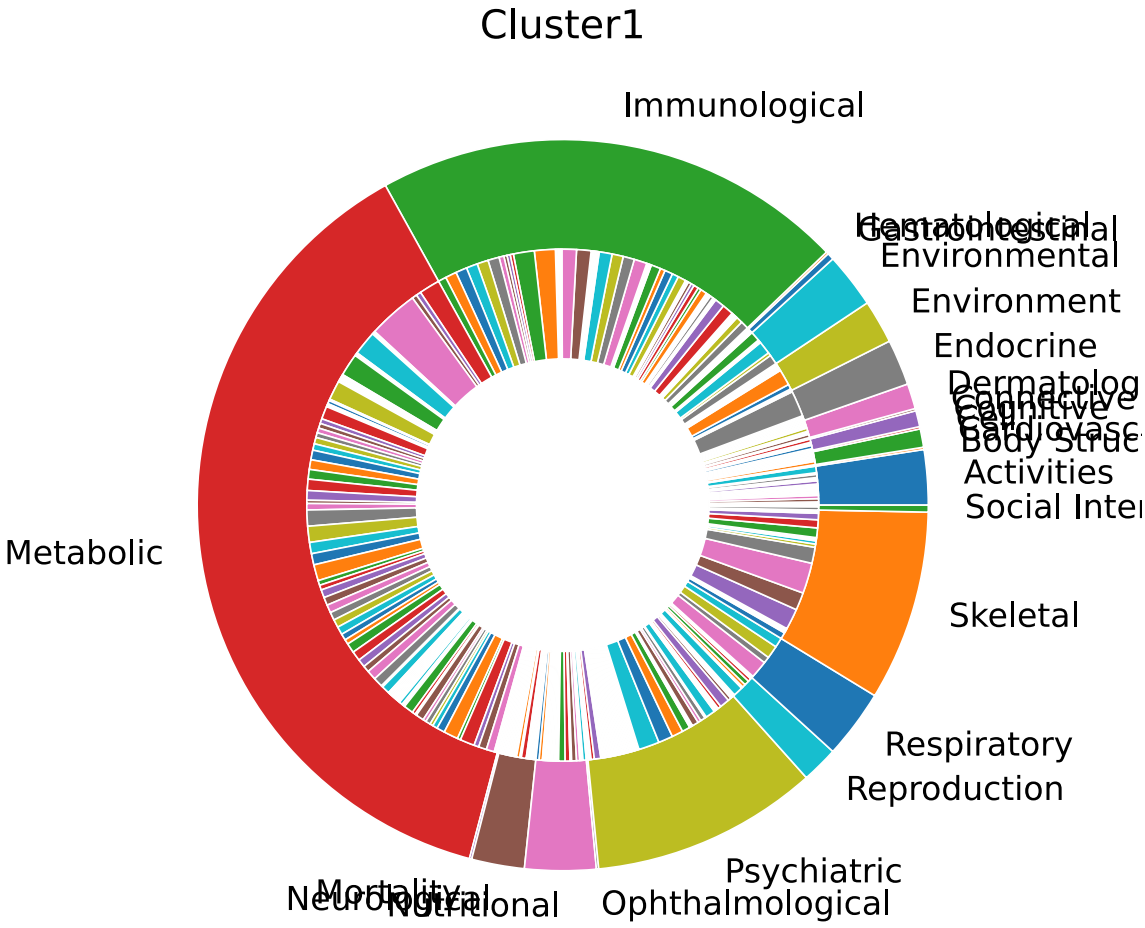


该图通过簇标准化展现了簇内不同性状之间的差异，对各簇来说代谢、免疫相关的信号都较强

簇1





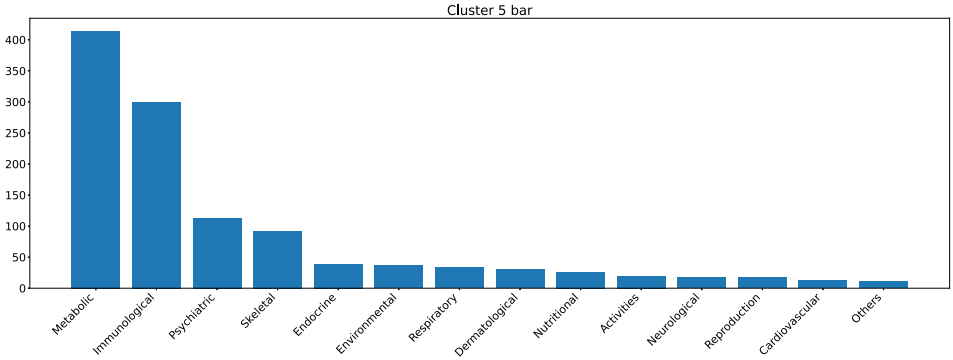


观察热图，发现簇1在环境形状方面较其他簇有较强信号，其数据如下

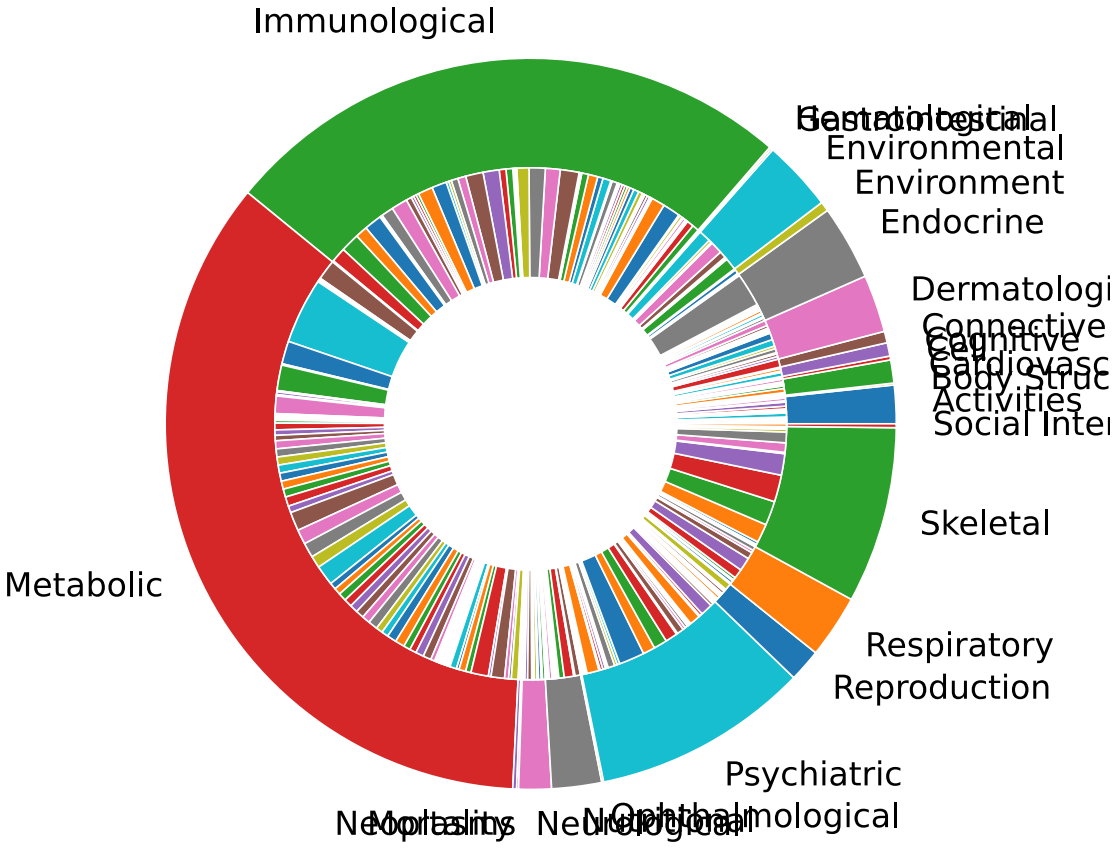
Field	Traits	Count
Environment	Educational attainment	10
Environment	Education - Qualifications	3
Environment	Attendance/disability/mobility allowance: Blue badge	1
Environment	Illnesses of mother: Diabetes	1
Environment	Illnesses of siblings: Diabetes	1
Environment	Illnesses of siblings: High blood pressure	1
Environment	Job involves heavy manual or physical work	1
Environment	Maternal smoking around birth	1

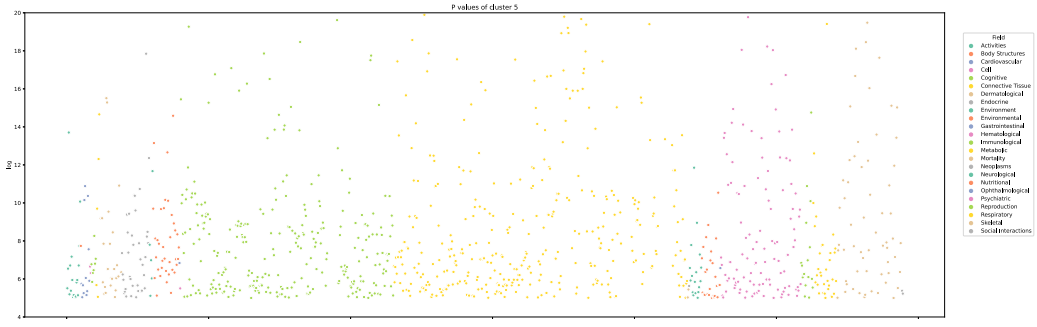
发现其中竟然与教育程度有关，推测是因为教育程度较低的个体后期从事体力劳动的可能性较大，对关节有损伤，我们试图将该簇定义为外因组

簇5



Cluster5





簇5相对于其他簇各个性状域均有较强信号，其中以内分泌与免疫尤甚，分析其

Field	Traits	Count
Endocrine	Type 2 Diabetes	25
Endocrine	Diabetes (diagnosed by doctor)	2
Endocrine	Diagnoses - secondary ICD10: E11 Type 2 diabetes mellitus	2
Endocrine	Non-cancer illness code, self-reported: diabetes	2
Endocrine	Diagnoses - secondary ICD10: E03 Other hypothyroidism	1
Endocrine	Free thyroxine (FT4)	1
Endocrine	Non-cancer illness code, self-reported: hypothyroidism/myxoedema	1
Endocrine	Thyroid-stimulating hormone	1
Endocrine	Thyroid-stimulating hormone (female)	1
Endocrine	Thyroid-stimulating hormone (male)	1
Endocrine	Type 1 Diabetes	1
Endocrine	Type 2 Diabetes (adjusted for BMI)	1

Field	Traits	Count
Immunological	Myeloid white cell count (three-way meta)	14
Immunological	White blood cell count (three-way meta)	14
Immunological	Granulocyte count (three-way meta)	13
Immunological	Platelet distribution width (two-way meta)	13
Immunological	Sum neutrophil eosinophil count (three-way meta)	13
Immunological	Neutrophil count (three-way meta)	12
Immunological	Platelet distribution width (three-way meta)	12
Immunological	Sum basophil neutrophil count (three-way meta)	12
Immunological	Myeloid white cell count (two-way meta)	11
Immunological	Red cell distribution width (three-way meta)	11
Immunological	Red cell distribution width (two-way meta)	11
Immunological	White blood cell count (two-way meta)	11

Field	Traits	Count
Immunological	Granulocyte count (two-way meta)	10

我们发现骨关节炎与二型糖尿病以及与巨噬细胞数量的强烈关联，推测此组主要通过自体免疫的方式影响骨关节炎进程；我们将其定义为内因组