

Genome wide association study of silent cerebral infarction in sickle cell disease (HbSS and HbSC)

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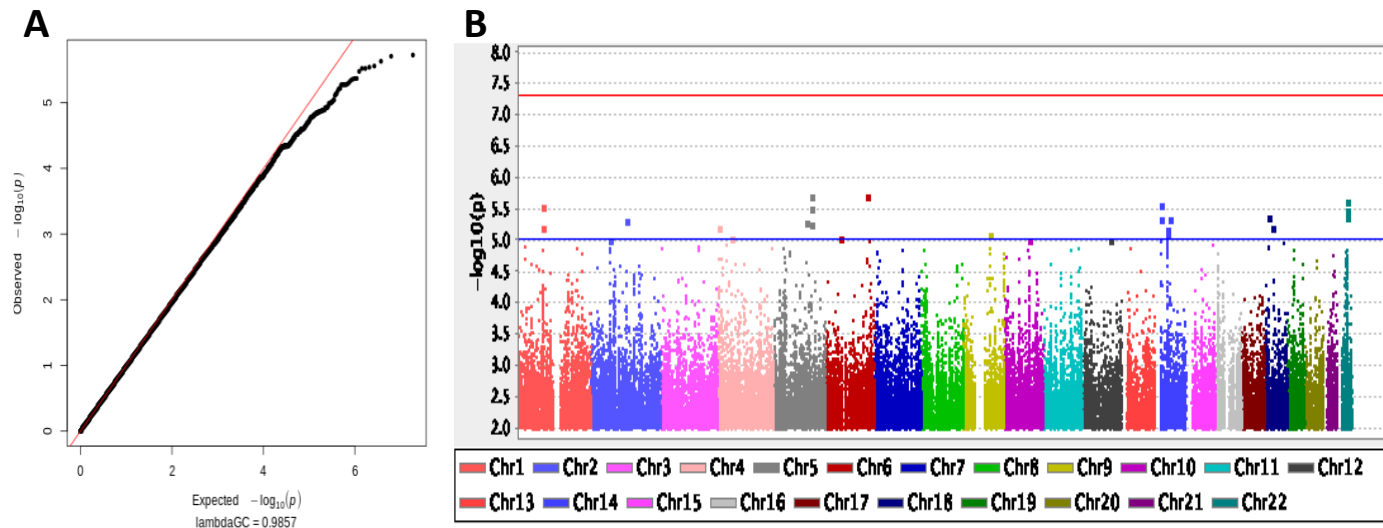
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Supplementary Figure 1: Results of genome wide association testing of SCI outcomes in patients with sickle cell disease. Age, gender, sickle genotype and alpha thalassemia were used as covariates, and a genetic relatedness included to control for population structure. The QQ plot (A) demonstrates no genomic inflation which would be suggestive of population stratification bias. The Manhattan plot (B) demonstrates the p values of the variants analysed. The blue line represents 1×10^{-5} , whilst the red line represents 5×10^{-8} which was used as the threshold of statistical significance.



Supplementary table 1 The top five variants identified in the GWAS described in supplementary figure 1.

| Gene | rsID | Change | MAF | OR | p | Variant location | Gene Function |
|---------|-------------|--------|-------|------|----------|------------------|---|
| CHSY3 | rs1557759 | G>A | 0.222 | 0.44 | 1.88E-06 | Upstream variant | Glycosyltransferase |
| PHACTR2 | rs6930487 | G>A | 0.388 | 1.94 | 1.96E-06 | Intronic | Platelet response to cytosolic Ca^{2+} |
| None | rs201658643 | G>GTA | 0.318 | 2.08 | 2.34E-06 | Intergenic | NA |
| TOX4 | rs10142478 | A>C | 0.067 | 0.25 | 2.76E-06 | Intronic | Chromatin Binding |
| PRKACB | rs2250806 | A>G | 0.415 | 0.51 | 2.88E-06 | Intronic | Mediates cAMP-dependent signalling |