Global transcriptome and chromatin occupancy analysis reveal the short isoform of GATA1 is deficient for erythroid specification and gene expression.

- Germline GATA1 mutations
  - Subset of patients with Black Diamond Blackfan anemia
  - Defective erythropoiesis

- GATA1 role?
  - GATA1 loss of expression
  - GATA1s expression

- Global gene expression
  - Erythroid gene expression program activation
  - Terminal differentiation

- Chromatin occupancy analysis
  - Binding efficiency
    - GATA1 erythroid-specific target genes

Chlon et al., Haematologica, 2015