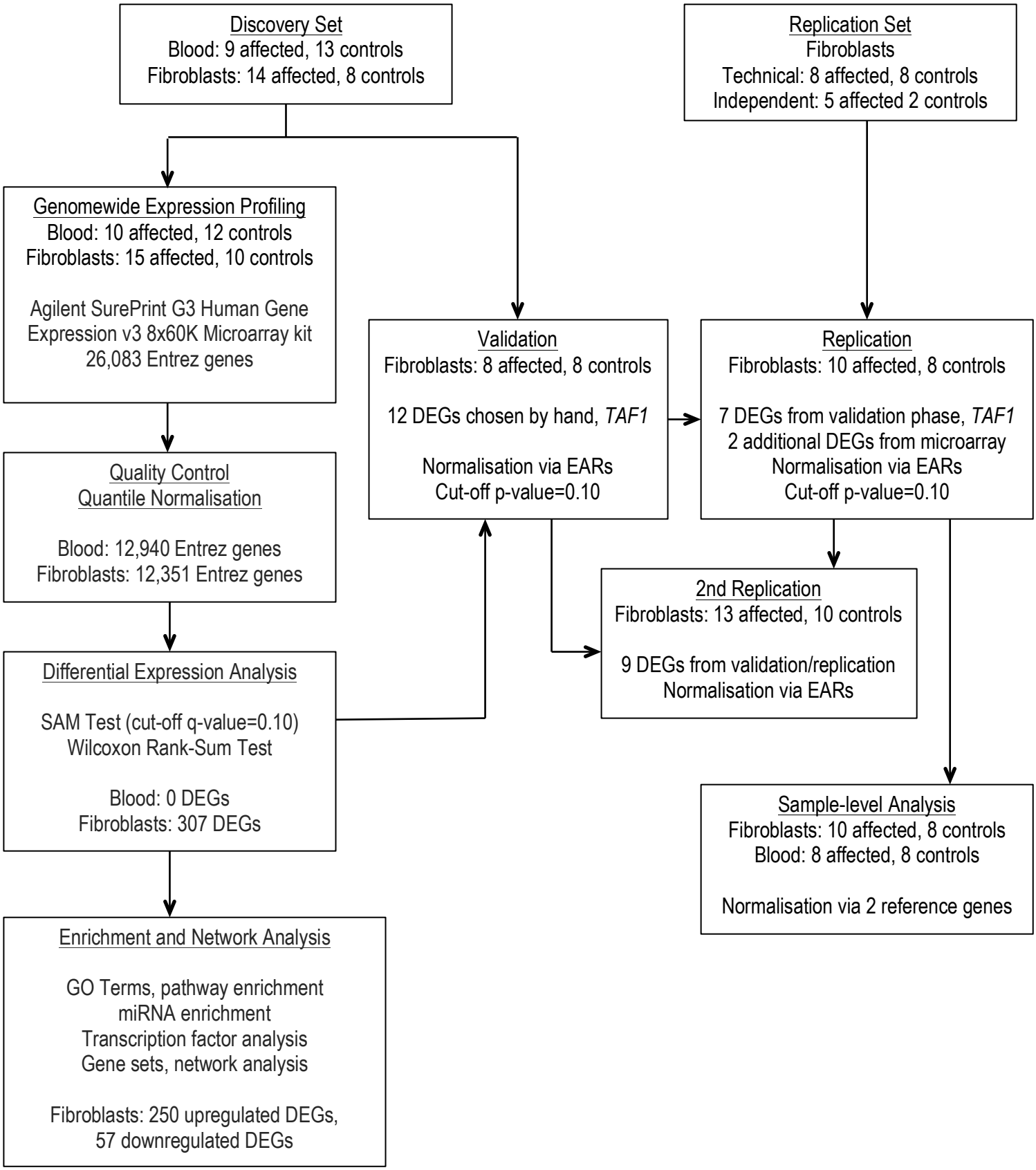
Discovery Set
Blood: 9 affected, 13 controls
Fibroblasts: 14 affected, 8 controls
Replication Set
Fibroblasts
Technical: 8 affected, 8 controls
Independent: 5 affected 2 controls
Genomewide Expression Profiling
Blood: 10 affected, 12 controls
Fibroblasts: 15 affected, 10 controls
Agilent SurePrint G3 Human Gene Expression v3 8x60K Microarray kit
26,083 Entrez genes
Validation
Fibroblasts: 8 affected, 8 controls
12 DEGs chosen by hand, TAF1
Normalisation via EARs
Cut-off p-value=0.10
Replication
Fibroblasts: 10 affected, 8 controls
7 DEGs from validation phase, TAF1
2 additional DEGs from microarray
Normalisation via EARs
Cut-off p-value=0.10
Quality Control
Quantile Normalisation
Blood: 12,940 Entrez genes
Fibroblasts: 12,351 Entrez genes
2nd Replication
Fibroblasts: 13 affected, 10 controls
9 DEGs from validation/replication
Normalisation via EARs
Differential Expression Analysis
SAM Test (cut-off q-value=0.10)
Wilcoxon Rank-Sum Test
Blood: 0 DEGs
Fibroblasts: 307 DEGs
Sample-level Analysis
Fibroblasts: 10 affected, 8 controls
Blood: 8 affected, 8 controls
Normalisation via 2 reference genes
Enrichment and Network Analysis
GO Terms, pathway enrichment
miRNA enrichment
Transcription factor analysis
Gene sets, network analysis
Fibroblasts: 250 upregulated DEGs,
57 downregulated DEGs