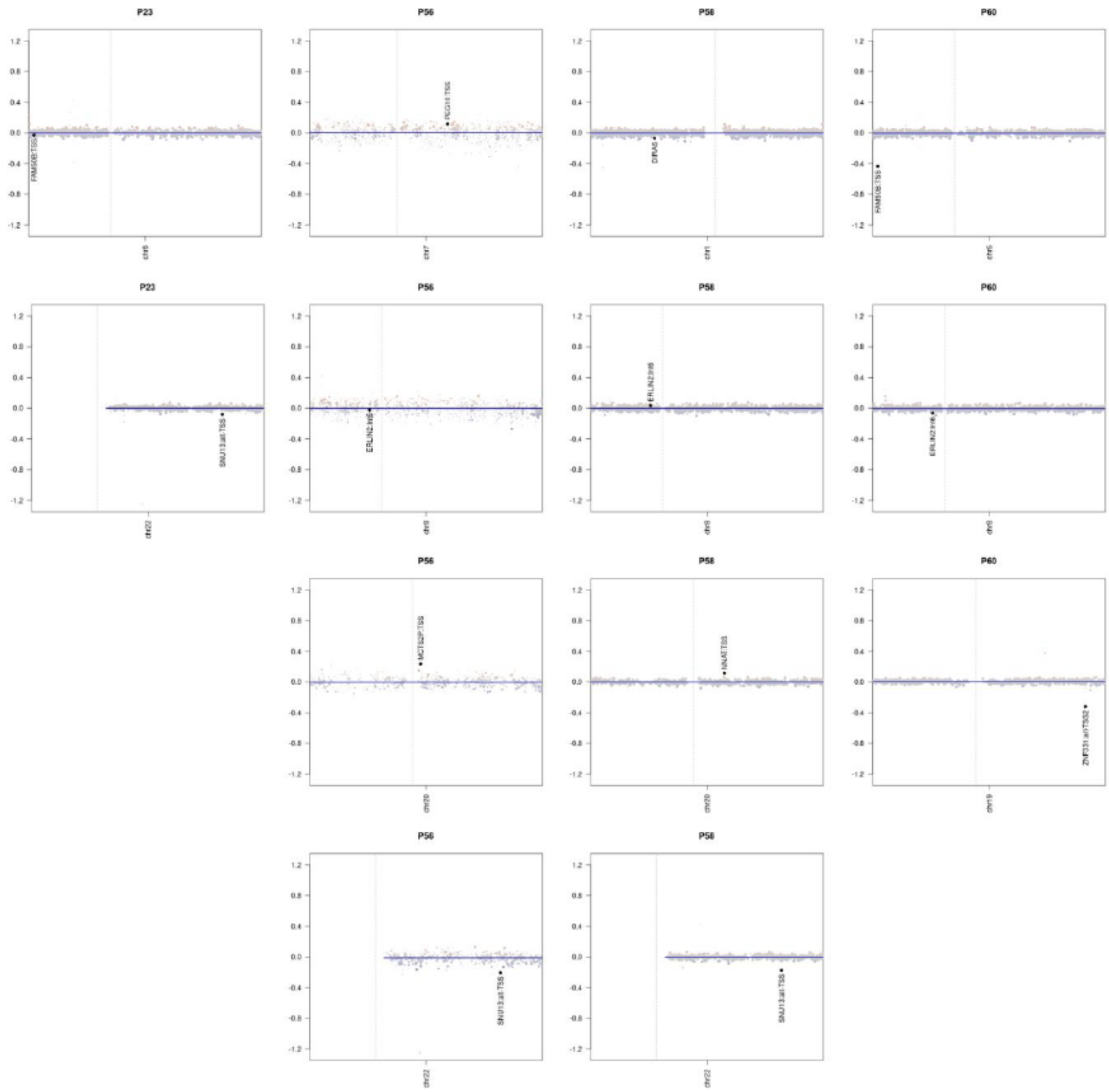

Fig. S1 Plot displaying the CNV status of the chromosome regions containing the iDMRs with abnormal methylation identified by EPIC array analysis in the patients 23, 56, 58 and 60.

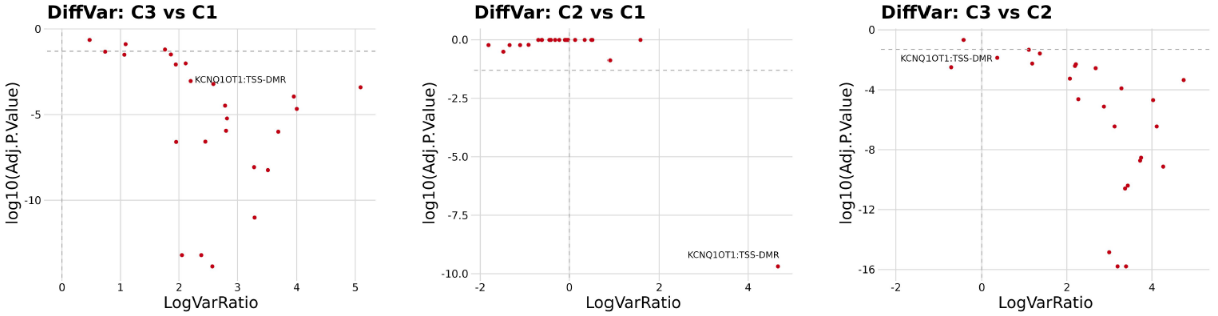

Fig. S2 Scatterplots displaying the differentially variable iDMRs between the C3 patient group and controls (C1, left panel), C2 patient group and controls (middle panel) and the C3 and C2 patient groups (right panel). The significance (log10(adj.p.value)) is indicated on the Y-axis, fold difference (LogVarRatio) on the X-axis.

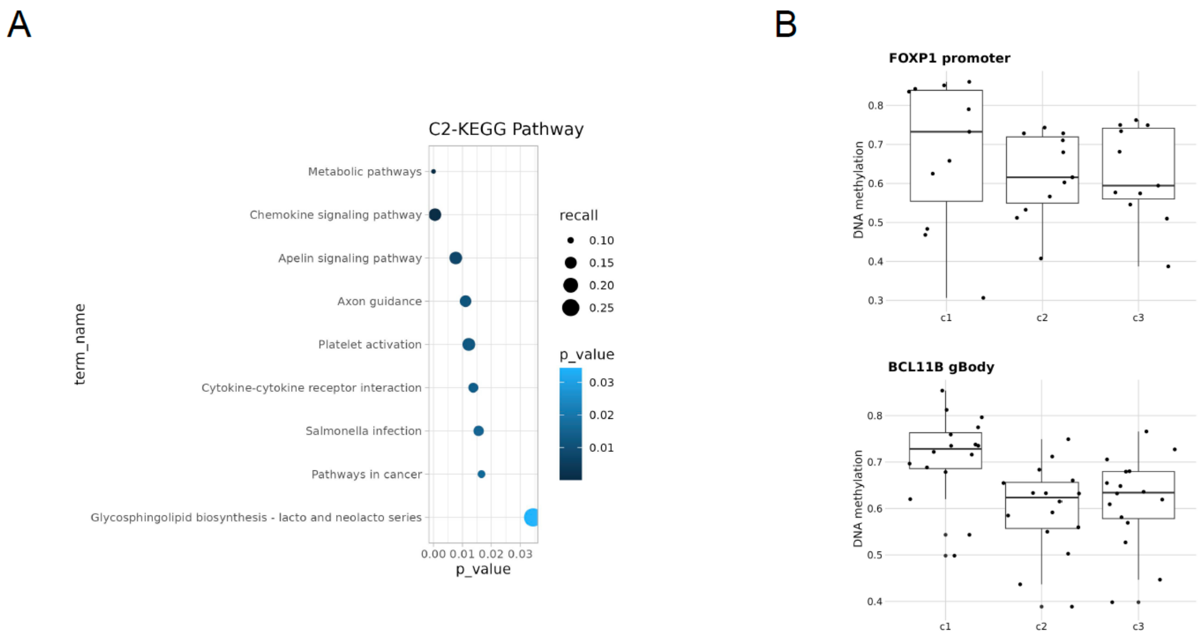

Fig. S3 Pathways and genes associated with DMPs. **A** KEGG pathway enrichment analysis of genes associated with DVPs in the C2 patient group, represented as in Fig. 4C. **B** Boxplot showing the methylation level (beta-values) of the FOXP1 promoter and BCL11B gene body in the C2 and C3 patient groups and controls (C1). Each dot represents a CpG.

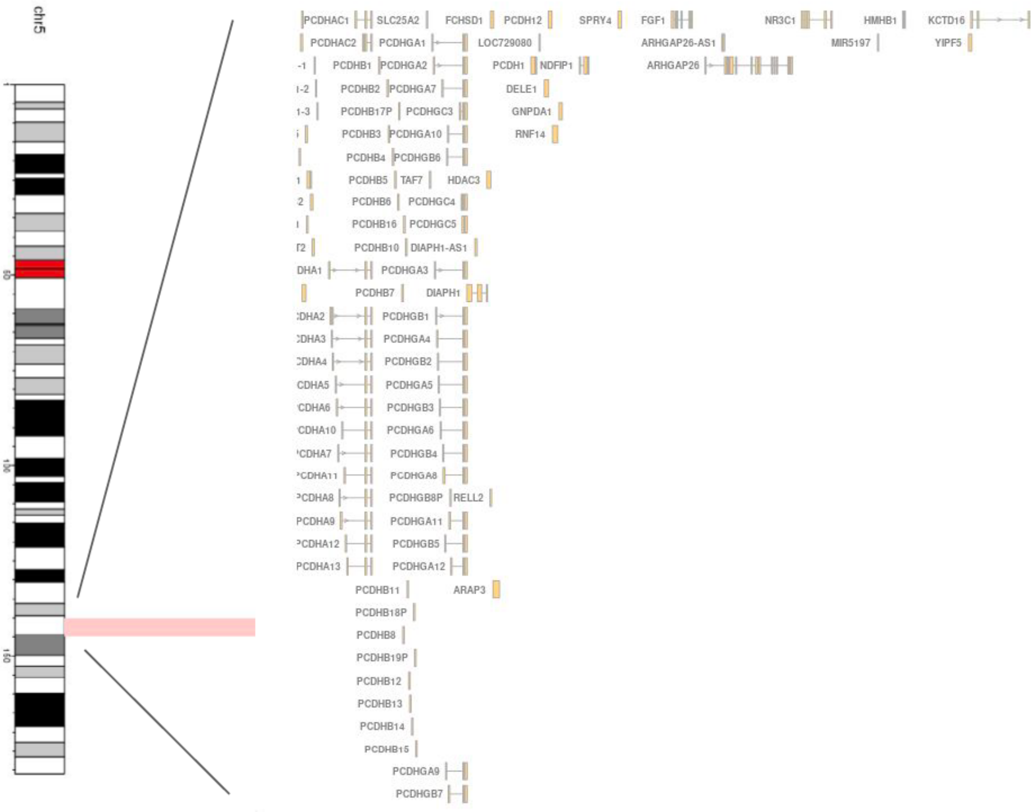

Fig. S4 Ideogram representing the chr5q31.3 band and the genes affected in by DVPs in the C3 patient group.

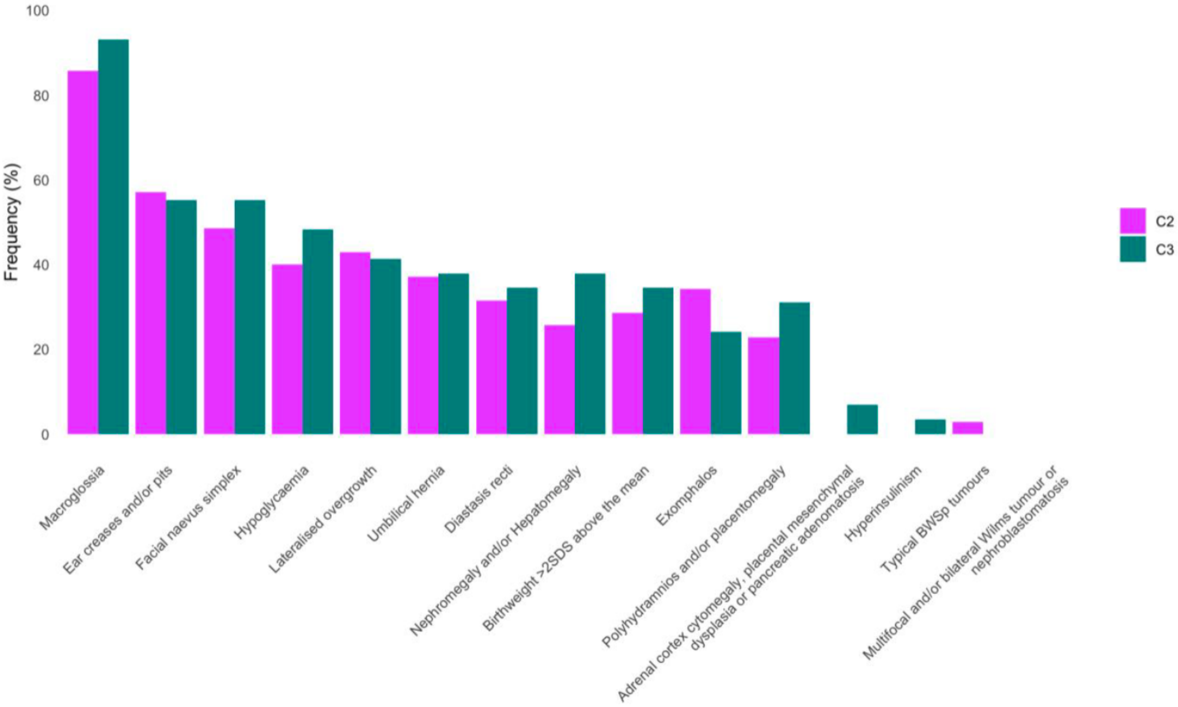

Fig. S5 Frequency of BWS clinical features in C2 (pink) and C3 (green) patient groups.