Abbreviations

ASE	allele-specific expression
ASW	African ancestry in Southwest USA
BMI	body mass index
bp	base pairs
c-dSNP	causal disease SNP
c-eQTL	causal eQTL SNP
CD	Crohn's disease
cDNA	copy DNA
CEPH	Centre d'Étude du Polymorphisme Humain
CEU	Utah residents with Northern of Western European ancestry
CHB	Han Chinese in Beijing, China
CHD	Chinese in Metropolitan Denver, Colorado, USA
ChIP	chromatin immunoprecipitation
cM	centimorgan
CNV	copy number variant
cRNA	copy RNA
cSNP	causal SNP
dSNP	disease SNP
DZ	dizygotic
EBV	Epstein-Barr virus
ENCODE	encyclopedia of DNA elements
eQTL	expression quantitative trait locus
EST	expressed sequence tag
FA	factor analysis
FDR	false discovery rate
GIH	Gujarati Indians in Houston, Texas, USA
GWAS	genome-wide association study/studies
IVT	in vitro transcription
JPT	Japanese in Tokyo, Japan
kb	kilobase
KCL	King's College London
LCLs	lymphoblastoid cell lines (EBV-transformed B-cells)
LCR	locus control region
LD	linkage disequilibrium
LR	linear regression
LWK	Luhya in Webuye, Kenya
MAF	minor allele frequency
Mb	megabase
MCTA	matched co-twin analysis
MEMN	macrophage enriched metabolic network
MEX	Mexican ancestry in Los Angeles, California, USA

miRNA	microRNA
MKK	Maasai in Kinyawa, Kenya
MS	multiple sclerosis
MuTHER	multiple tissue human expression resource
MZ	monozygotic
NHGRI	National Human Genome Research Institute
PCA	principal component analysis
PT	permutation threshold
QC	quality control
QTL	quantitative trait locus
RNA-seq	RNA sequencing
RTC	regulatory trait concordance
SAM	sentrix array matrix
SNP	single nucleotide polymorphism
SRC	Spearman rank correlation
SSAHA	sequence search and alignment by hashing algorithm
T2D	type 2 diabetes
TF	transcription factor
TP	true positives
TSI	Toscans in Italy
TSS	transcription start site
UGMS	University of Geneva Medical School
UTR	untranslated region
WTCCC	Wellcome Trust Case Control Consortium
WTCHG	Wellcome Trust Centre for Human Genetics
WTSI	Wellcome Trust Sanger Institute
YRI	Yoruban in Ibadan, Nigeria

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Appendix

1. **Biopsy technique protocol**

1. The lower abdominal biopsy site is cleaned. Local anesthetic with adrenaline is infiltrated into the pre-inked skin.

2. Stretching the skin perpendicular to the relaxed skin tension lines between thumb and finger either side of the area to be sampled, the punch blade is placed on the skin and rotated under gentle pressure by rolling it between the thumb and finger using a twisting drilling action.

3. One should penetrate to the level of the fat layer to achieve a full thickness skin biopsy specimen. The specimen should be weighed, cut in half, and stored immediately in liquid nitrogen

4. The specimen will either float up on the fat layer or can be gently lifted using a skin hook or gently applied forceps to allow specimen collection by cutting through the fat layer using a scalpel or sharp scissors.

5. Further fat can be obtained by careful dissection of the fat layer using forceps and scalpel. The fat sample should be weighed and immediately stored in liquid nitrogen.

6. Haemostasis can be achieved with direct pressure and/or interrupted sutures. Both absorbent and non-absorbent sutures can be used in a layered closure for larger punch defects.

7. The resultant defect can be allowed to heal by secondary intention as an alternative method although optimal haemostasis and cosmesis as well as reduced healing time are usually seen with sutured wounds.