



Almac Diagnostic Services

Chronic Disease Case Studies

March 2024



Chronic Disease Indications



RENAL



LIVER



RESPIRATORY



BRAIN



IBD



AUTOIMMUNE



ALLERGY



CARDIOVASCULAR



ENDOCRINE



INFECTIOUS DISEASE



METABOLIC



OCULAR

Almac Chronic Disease Programmes

RENAL



LIVER



RESPIRATORY



AUTOIMMUNE



ALLERGY



INFLAMMATORY BOWEL DISEASE



BRAIN



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INFLAMMATORY BOWEL DISEASE



BRAIN



RENAL

CASE STUDY # 1

Background: Ongoing CDx Programme with large global pharma partner

Objective: To develop and validate a clinical trial assay (CTA) to determine eligibility for treatment with a targeted therapeutic. IVD CDx kitted assay in future.

Indication: Chronic kidney disease (CKD)

Diagnostic Gene Focus: Germline variant

Platform / Technology: qPCR - ThermoFisher QuantStudio5Dx

Sample Type: Whole blood and buccal swab

Data Sciences & Reporting: Customised trial reporting software development & validation for client



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LIVER

CASE STUDY # 1

Background: Ongoing IVD Kit CDx Programme with large global pharma partner

Objective: To develop and validate a clinical trial assay (CTA) to determine eligibility for treatment with a targeted therapeutic. IVD CDx kitted assay in future.

Indication: Chronic liver disease (NASH / MASH)

Diagnostic Gene Focus: Germline variant

Platform / Technology: qPCR - ThermoFisher QuantStudio5 Dx

Sample Type: Whole blood and buccal swab

Data Sciences & Reporting: Custom trial reporting software developed & validation



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CASE STUDY # 1

Background: Ongoing IVD Kit CDx Programme with large global pharma partner

Objective: To develop and validate a clinical trial assay (CTA) to determine eligibility for treatment with a monoclonal antibody. IVD CDx kitted assay in future.

Indication: Asthma (Moderate to Severe)

Diagnostic Gene Focus: Multiple germline polymorphisms in selected genes

Platform / Technology: NGS - Illumina MiSeq

Sample Type: Whole blood

Data Sciences & Reporting: Development of a germline variant calling pipeline & custom reporting software



RESPIRATORY

CASE STUDY # 2

Background: Completed project with Biopharma client to develop an RUO NGS assay to target an asthma drug

Objective: To perform Total RNAseq on pre-treatment blood samples & perform biomarker signature development. Data generated by Almac from the retrospective testing will be used to identify a predictive biomarker for client's drug clinical trial

Indication: Asthma (Severe)

Diagnostic Gene Focus: Multiplex gene signature developed

Platform / Technology: NGS - Illumina NovaSeq

Sample Type: Blood

Data Sciences & Reporting: Bespoke bioinformatic analyses



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CASE STUDY # 3

Background: Completed project with Biopharma client to develop a Microarray based predictive biomarker for COPD drug

Objective: To profile matched induced sputum and blood samples on microarray and perform biomarker signature development. Data generated by Almac from the retrospective testing will be used to identify a predictive biomarker for client's phase 2a clinical trial

Indication: Chronic Obstructive Pulmonary Disease (COPD)

Diagnostic Gene Focus: Gene expression signature development

Platform / Technology: Gene expression using Affymetrix Gene Chip Microarray

Sample Type: Induced sputum and blood

Data Sciences & Reporting: Customised reporting for client. Additional bioinformatics analysis on dataset performed (differential gene expression and functional enrichment analysis) identified a group of sub-responders. Two manuscripts published



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CASE STUDY # 1

Background: Completed RUO panel assay for use in a retrospective clinical trial for global biopharma client for lupus drug development

Objective: To develop and validate a multiple gene qPCR (RUO) assay and perform retrospective testing of samples collected during a Ph2b clinical trial.
The aim was to evaluate the efficacy and safety of their drug in Systemic Lupus Erythematosus (SLE)

Indication: Lupus (SLE)

Diagnostic Gene Focus: Expression of multiple target genes & additional housekeeping genes

Platform / Technology: qPCR - Fluidigm HD BioMark system

Sample Type: Whole blood

Data Sciences & Reporting: Biostatistics team developed an automated data analysis tool to reduce timelines to perform data analysis



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CASE STUDY # 1

Background: Ongoing project with Global Pharma client to develop an RUO qPCR assay

Objective: RUO qPCR clinical trial assay utilised in a phase 3 drug study. Non interventional testing complete. Interventional study: TBC

Indication: Severe Allergy

Diagnostic Gene Focus: Multiple germline SNPs across selected genes that predispose to allergy

Platform / Technology: qPCR - ThermoFisher QuantStudioDx

Sample Type: Whole blood

Data Sciences & Reporting: Custom trial reporting software developed & validated



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CASE STUDY # 1

Background: Assisted Global Pharma client to develop a qPCR signature to predict response to α monoclonal antibody
Potential to now assist with assay development & validation of the Non-Response Signature as a CTA for prospective use in clinical trials

Objective: Hypothesis: Could the signature be used as a negative predictor clinical trial stratification tool and future CDx?

Indication: Ulcerative Colitis (UC)

Diagnostic Gene Focus: qPCR platform for expression of multiple selected genes

Platform / Technology: qPCR - QuantStudio Dx

Sample Type: Colon biopsies in RNALater®

Data Sciences & Reporting: Application of bespoke algorithm to produce assay score and response call



INFLAMMATORY BOWEL DISEASE

CASE STUDY # 2

Background: Almac partnered with Biotech company for Bioinformatics & Software services to help them to launch the world's first validated biomarker prognostic test in inflammatory mediated diseases

Objective: Development of a portable software solution for whole blood prognostic gene expression test

Indication: IBD (Crohn's Disease & Ulcerative Colitis)

Diagnostic Gene Focus: Multiple selected genes on a panel based approach

Platform / Technology: qPCR based assay for gene expression

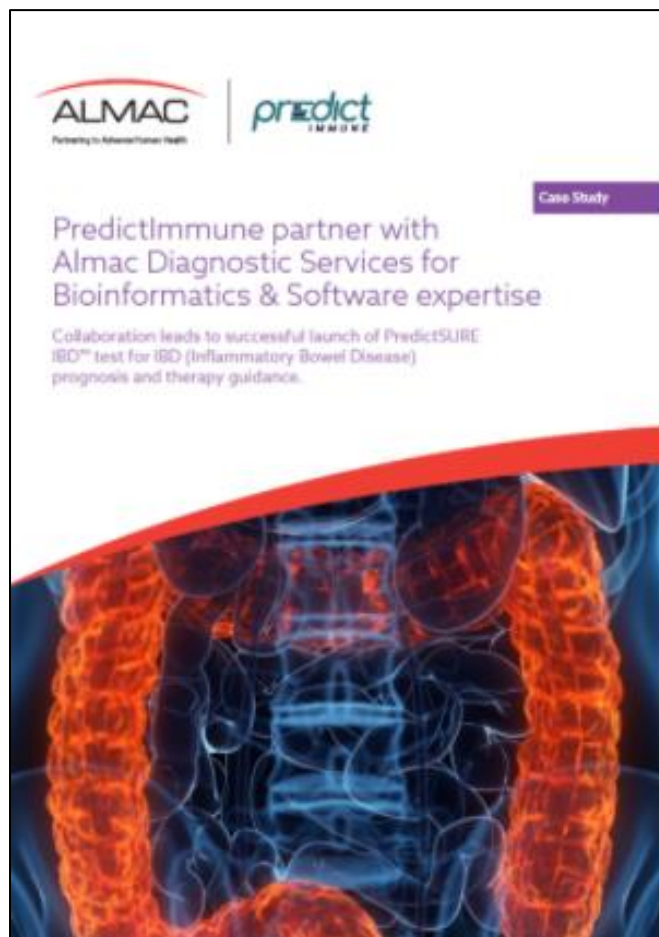
Sample Type: Whole blood

Data Sciences & Reporting: Bespoke bioinformatics support, software development & algorithm generation

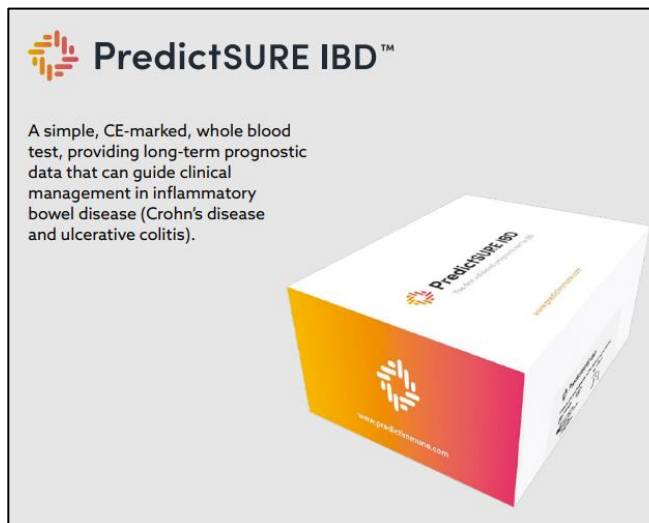




IBD - Bioinformatics & Software Support



“PredictImmune and Almac partnered to develop and validate the gene expression software algorithm used to interpret the results from the world’s first validated and CE-marked prognostic test for inflammatory bowel disease. PredictImmune selected Almac Diagnostic Services as their preferred provider for bioinformatics, software design and development solutions to support the successful commercialisation and launch of PredictSURE IBD™.”



To read the full case study:

[LINK](#)

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BIPOLAR DISORDER

CASE STUDY #1

Background: Ongoing testing to support a clinical trial sponsored by a biopharma client to assess the safety and efficacy of an investigational product for the treatment of a neuropsychiatric disorder

Objective: Single nucleotide polymorphism (SNP) genotyping in a prospective setting using a qPCR genotyping assay. Data generated by Almac will be used to determine the eligibility for enrolment for client's phase 2a clinical trial

Indication: Bipolar Disorder

Diagnostic Gene Focus: Identifying gain of function and loss of function single nucleotide polymorphisms (SNPs) in selected gene that might predict response to the drug

Platform / Technology: qPCR - Quantstudio 5 Dx

Sample Type: Whole blood

Data Sciences & Reporting: Customised reporting software developed for client. Monthly production of file transfers and weekly informal file transfers





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