

One-stop Solution for Rare Disease Diagnostic Development



Contact Us



info@protheragen.us



1-631-533-2057



diagnostics.protheragen.us



101-4 Colin Dr, Holbrook, NY 11741, USA





Overview

About Protheragen	3
Rare Diseases in Focus	4
One-stop Solutions	5
IVD Reagent/Kit Development Service	6
IVD Device Development Service	8
Point-of-Care Testing (POCT) Development Service	9
Companion Diagnostic Development Service	12
Collaboration Opportunities	13



// About Protheragen

As a leading provider of preclinical research services, **Protheragen** specializes in delivering end-to-end *in vitro* diagnostic (IVD) development services for rare diseases. Our comprehensive solutions encompass the entire workflow from biomarker discovery, assay development to preclinical validation, supporting our clients in transitioning from diagnostic research to commercialization seamlessly.

Customized Solutions



- IVD Reagent/Kit Development
- IVD Device Development
- Point-of-Care Test (POCT) Development
- Companion Diagnostic Development

Our Advantages



Innovative
Platforms



Rich
Expertise



Customized
Solutions



Trusted
Partner



Cutting-edge Diagnostic Research on Multiple Rare Diseases

Rare diseases, also known as orphan diseases, are characterized by low incidence in the population. Despite their rarity, more than 7,000 rare diseases have been discovered, affecting millions of patients worldwide. The diagnosis of rare diseases faces great challenges due to their diversity and complex clinical manifestations, resulting in many patients being delayed or misdiagnosed. Focusing on precision and innovation, **Protheragen** is committed to providing cutting-edge diagnostic development solutions for a variety of rare diseases to simplify diagnostic procedures and revolutionize the diagnostic landscape.



Genetic
Diseases



Infectious
Diseases



Autoimmune
Diseases



Metabolic
Diseases

Here are some examples of rare diseases for our focus:

- Cystic Fibrosis (CF)
- Hepatitis B
- Hemophilia
- Phenylketonuria (PKU)
- Multiple Sclerosis (MS)
- Duchenne Muscular Dystrophy (DMD)
- Spinal Muscular Atrophy (SMA)
- Coronavirus Disease 2019 (COVID-19)
- Hand, Foot, and Mouth Disease (HFMD)
- Respiratory Syncytial Virus (RSV) Infection
- Rheumatoid Arthritis (RA)
- Systemic Lupus Erythematosus (SLE)
- Isovaleric Acidemia (IVA)
- Lysosomal Storage Diseases (LSDs)
- Others

Protheragen specializes in diagnostic development for rare diseases, offering a comprehensive one-stop solution that includes IVD reagents/kits, IVD equipment, point-of-care testing (POCT), and companion diagnostic development services. We adhere to stringent development processes, ensuring high-quality and reliable solutions that meet regulatory standards and cater to the unique needs of our clients in the field of rare disease diagnostics.



IVD Reagent/Kit Development Service

To comprehensively address diverse diagnostic needs in the field of rare diseases, we focus on developing a variety of types of IVD reagents/kits, including antigen, antibody, nucleic acid, metabolite, and microbial detection products.



IVD Device Development Service

Protheragen is dedicated to the design and development of advanced IVD devices. From concept development to prototype manufacturing, our team ensures these devices meet the highest standards of performance and usability.



Point-of-Care Testing (POCT) Development Service

Specializing in POCT development, we prioritize the creation of rapid and precise diagnostic solutions. By delivering swift and accurate results, we facilitate prompt decision-making in resource-constrained regions and enable home-based testing.



Companion Diagnostic Development Service

Companion diagnostics can identify specific biomarkers to help tailor personalized treatment plans for patients. Protheragen focuses on the development of companion diagnostics that meet the latest scientific advances and regulatory standards.

Service Workflow

At **Protheragen**, our IVD reagent/kit development services follow a rigorous process that covers concept validation and feasibility assessment, IVD reagent/kit design, validation and optimization. We combine cutting-edge technologies with rigorous testing and optimization procedures to provide high-quality IVD reagents and kits to meet the unique needs of our customers in the field of rare disease diagnosis.



Proof of
Concept and
Feasibility



IVD
Reagent/Kit
Design



IVD
Reagent/Kit
Verification



IVD
Reagent/Kit
Optimization



Antigen Detection Reagents/Kits: Used for detection of pathogen surface proteins, tumor biomarkers or toxins.



Antibody Detection Reagents/Kits: Used for detection of pathogen-specific antibodies, autoimmune antibodies and allergen-specific IgE.



Nucleic Acid Detection Reagents/Kits: Used for detection of genomic or transcriptome material (DNA/RNA) in pathogens or human cells.



Metabolite Detection Reagents/Kits: Used for the detection of metabolic intermediates, hormones or drugs in biological samples.



Microbial Detection Reagents/Kits: Used for detection of microorganisms, including bacteria, fungi, and parasites.

Protheragen specializes in developing IVD reagents/kits for four critical rare disease categories: genetic disorders, infectious diseases, autoimmune conditions, and metabolic diseases. Our solutions leverage advanced technologies such as PCR, immunoassays and biochemical analysis to target disease-specific biomarkers. These IVD products enable earlier detection, accurate subtyping and personalized treatment monitoring of diseases that are often misdiagnosed or missed in traditional medical settings.

Kits	Detection Methods	Targeted Diseases
CFTR Mutation Test Kit	ARMS-PCR	Cystic Fibrosis (CF)
MLPA DMD Kit	MLPA	Duchenne Muscular Dystrophy (DMD)
GAA Activity Assay Kit	Fluorometric Assay	Pompe Disease
SMN1/SMN2 Copy Number Kit	Digital PCR	Spinal Muscular Atrophy (SMA)
COVID-19 Antigen Test Kits	Lateral Flow Immunoassay	COVID-19
HBV DNA Detection Kit	Real-Time PCR	Hepatitis B (HBV)
Rheumatoid Factor IgM ELISA Kit	ELISA	Rheumatoid Arthritis (RA)
Antinuclear Antibody (ANA) Screen Kit	ELISA	Systemic Lupus Erythematosus (SLE)
Glycosaminoglycans (GAGs) Test Kit	Sandwich ELISA	Mucopolysaccharidoses (MPS)
Galactose Oxidase Assay Kit	Enzymatic Colorimetry	Galactosemia

Genetic Diseases

Infectious Diseases

Autoimmune Diseases

Metabolic Diseases

For more information about our [IVD reagent/kit](#) development services for [rare diseases](#), please contact us.

The advancement of IVD devices is crucial for the early detection and precise diagnosis of rare diseases. Our multidisciplinary team, comprising scientists, engineers, and regulatory experts, combines deep technical expertise with a passion for innovation. They are dedicated to providing high-performance and compliant solutions for the development of cutting-edge IVD devices. **Protheragen** employs a rigorous phased development process that guides projects from concept to commercialization.

Preparation of Device Development Plan

Based on the intended use of your IVD devices and the target rare diseases, we will first develop a comprehensive development roadmap. Our plan integrates user requirements, regulatory pathways and milestone timelines to ensure alignment with commercial objectives.

Styling Machine Development Service

Protheragen designs devices that align with ergonomic and aesthetic principles to seamlessly integrate into laboratories or clinics. By integrating industrial design, material selection and brand image, we develop high-quality devices that meet relevant standards.

01



03

02

Prototype Design Service

Drawing on expertise in CAD modeling, microfluidic engineering, and integrated analysis, we transform concepts into functional prototypes. Our designs prioritize usability, scalability, and detection performance. Rapid prototyping accelerates the iteration cycle.

Device Verification and Optimization Service

Through performance testing and experimental design analysis, we validate the sensitivity, specificity, and reproducibility of the devices. Pressure testing ensures stability, and data-driven improvements help prepare for clinical trials and regulatory submissions.

04

Customized POCT Product Development

- ◇ Point-of-care testing (POCT) refers to diagnostic tests that can be performed at or near the patient's location, providing immediate results without the need for laboratory infrastructure. POCT products mainly include rapid diagnostic kits and portable devices.
- ◇ Protheragen delivers end-to-end solutions for high-accuracy and user-friendly POCT products tailored to rare diseases.

LEADER IN IVD DEVELOPMENT

Product Name	Detection Technology/Application	Targeted Diseases
PKU Strip Test	Enzymatic Colorimetry	Phenylketonuria (PKU)
COVID-19 Antigen Rapid Test Kit	Lateral Flow Immunoassay	COVID-19
Hemoglobin S (HbS) Detection Kit	Solubility Test	Sickle Cell Disease
G6PD Rapid Diagnostic Test	Colorimetric Enzymatic Assay	G6PD Deficiency
MP IgM Rapid Test Kit	Rapid Immunochromatographic Assay	Mycoplasma Pneumoniae (MP) Infection
G6PD Analyzer	Determination of G6PD Enzyme Activity	G6PD Deficiency
Blood Gas Analyzer	Blood Gas/Electrolyte Analysis	Metabolic Diseases
Handheld Portable Analyzer	Acid-base/Blood Gas/Electrolyte Test	Metabolic Diseases or Genetic Disorders
Portable Blood Glucose Meter	Rapid Measurement of Blood Glucose Levels	Diabetes and Glycogen Storage Diseases

For more information about our POCT development services for rare diseases, please contact us.

Case Study-Development of POCT Platform Based on MetaSPR

Rapid and ultra-sensitive detection of small molecules is crucial for health management and biomarker detection in various diseases.

Protheragen has developed a chip based on metasurface plasmon resonance (MetaSPR) technology that can ultra-sensitively detect small molecules such as progesterone (P) and glucose (G) in the blood. This POCT platform holds great promise for applications in health management, early diagnosis, and other fields.

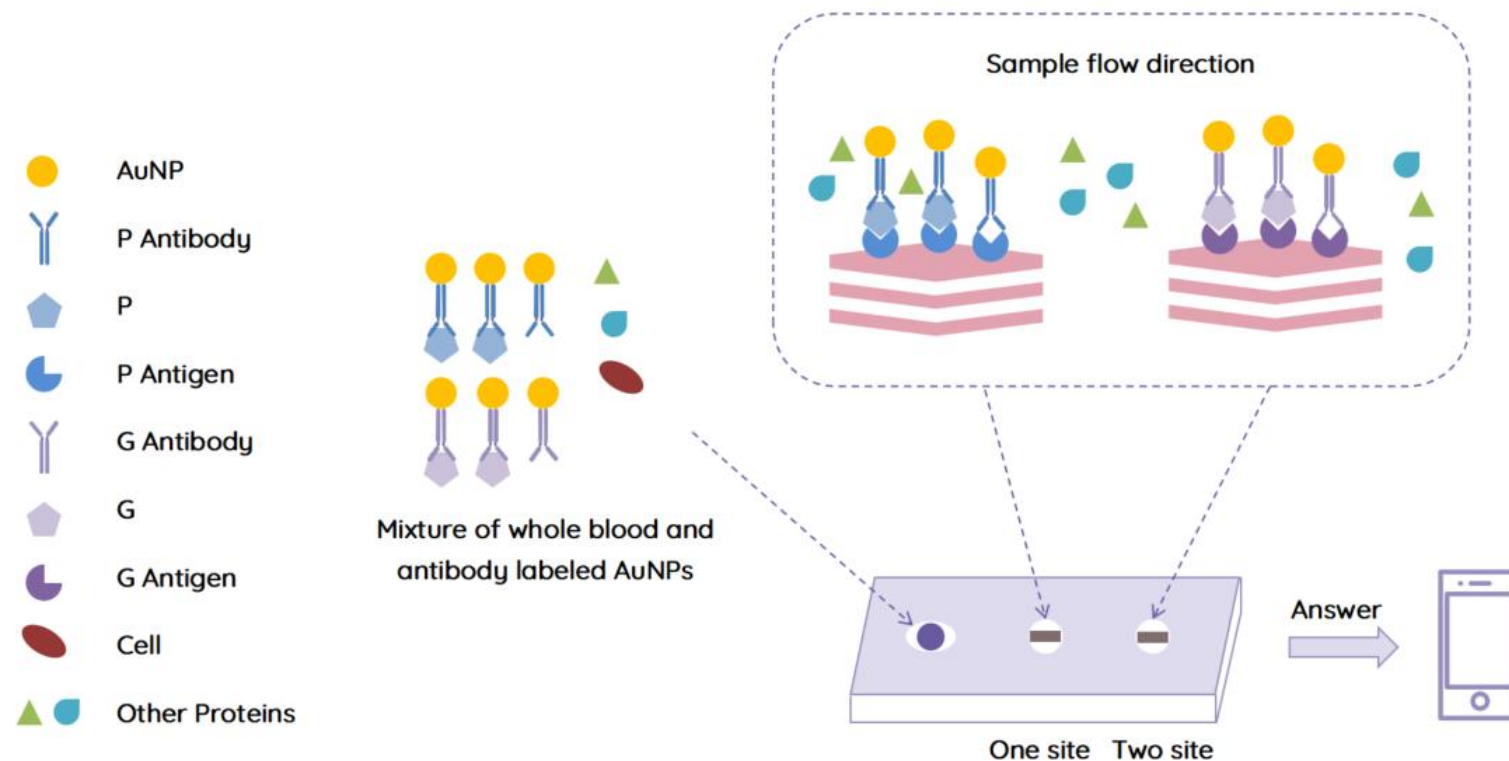


Fig.1 Multiplex small molecule testing controlled by smartphone on a POCT platform. This innovative POCT platform integrates MetaSPR chips with gold nanoparticle (AuNP) signal amplification, microlens-inspired light-homogenizing films, and microfluidic technology to enable ultra-sensitive, rapid, and portable detection of small molecules.

Case Study-Simultaneous Multiplexed Detection of Small Molecules

Using this POCT platform, Protheragen used competitive immunoassay technology for multiplexed small molecule detection (P and G) and recorded the signal with a smartphone. The results showed that simultaneous detection in buffer and whole blood was highly sensitive, showing a strong negative correlation between signal and concentration ($R^2 > 0.998$). The limit of detection (LOD) was improved by 1000 times compared to traditional methods, confirming the ultra-sensitivity, versatility and universality of this platform in early diagnosis.

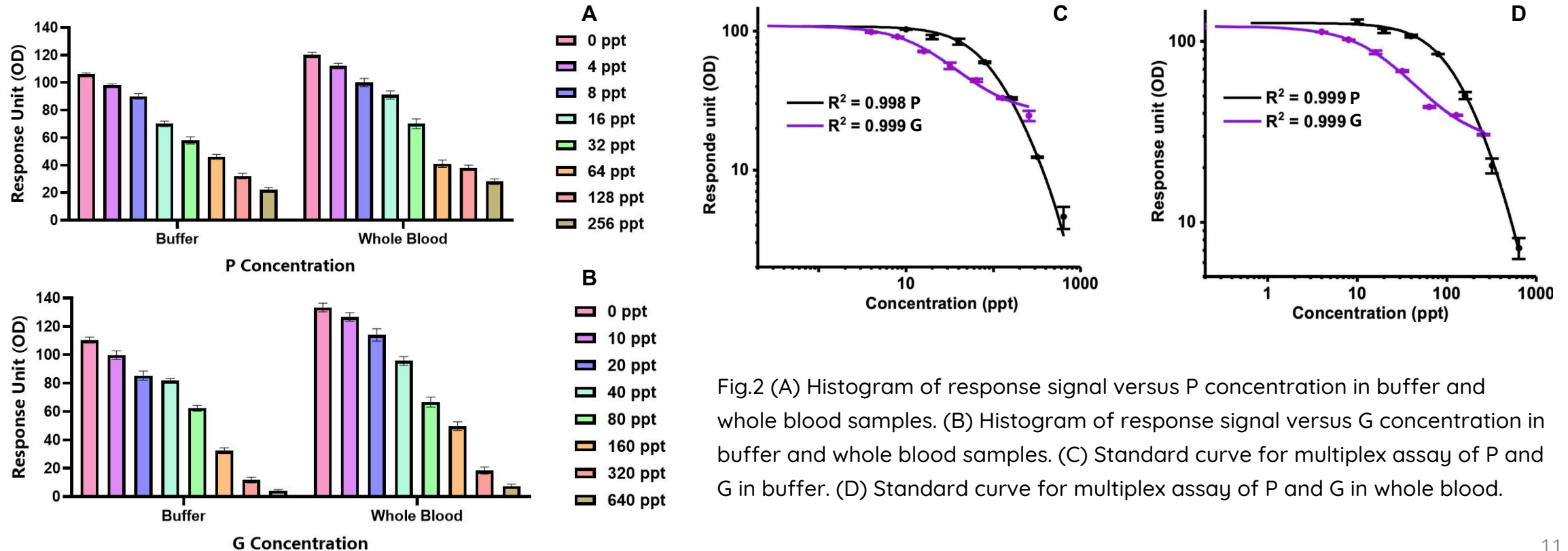


Fig.2 (A) Histogram of response signal versus P concentration in buffer and whole blood samples. (B) Histogram of response signal versus G concentration in buffer and whole blood samples. (C) Standard curve for multiplex assay of P and G in buffer. (D) Standard curve for multiplex assay of P and G in whole blood.

Companion diagnostics serve as a cornerstone of personalized medicine, customizing treatment strategies according to individual patients' distinct genetic profiles or specific biomarkers. **Protheragen** excels in crafting companion diagnostics during the preclinical phase, delivering comprehensive services for robust biomarker validation and assay optimization.



Biomarker Discovery & Validation

Protheragen uses NGS panels, mass spectrometry, and AI-driven bioinformatics to identify and qualify disease-specific biomarkers, with a focus on relevance for rare diseases. Our validation process includes sensitivity, specificity and repeatability assessments using clinically relevant samples and disease models to ensure the robustness of biomarkers in targeted therapies.



Assay Development & Optimization

Dedicated to developing cutting-edge diagnostic assays tailored for rare disease biomarkers, we employ a range of advanced technologies including PCR, NGS, and immunoassays. Throughout the entire optimization process, our primary focus lies in enhancing sensitivity, specificity, and robustness while customizing methodologies to effectively handle diverse and complex sample types.



Preclinical Testing

In the preclinical testing service, our scientists perform comprehensive analytical validation of diagnostic assays in a controlled laboratory environment following CLSI guidelines, including detection limit, precision, interference, and stability testing. This service produces a robust data package to support subsequent clinical development and regulatory submissions.



Accelerating Precision Diagnostics Through Strategic Partnerships

At **Protheragen**, we offer a comprehensive one-stop solution for rare disease diagnostic development, encompassing IVD reagents, kits, devices, POCT, and companion diagnostics. With deep expertise in rare disease diagnostics, we provide a full spectrum of customized services to support every stage of your diagnostic research journey. Let's collaborate to translate scientific insights into life-changing diagnostic approaches.



Expertise and Experience



Customized Solutions



Innovative Technology Platforms



Efficient and Reliable Partner

Your Trusted Partner in Rare Disease Diagnostic Development



Contact Us

✉ info@protheragen.us

☎ 1-631-533-2057

🌐 diagnostics.protheragen.us

📍 101-4 Colin Dr, Holbrook, NY 11741, USA

