提案人簡歷

張光遠 教授
資訊工程學系
電話：
02-2462-2192#6649
Email：
kchang@ntou.edu.tw

➢學經歷

- 美國聖路易華盛頓大學 計算生物學博士

➢研究專長

- 計算免疫學、生物資訊、健康資訊、人工智慧

➢代表著作

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➢ 曾獲獎項

- 指導學生獲 台海大資訊工程學系專題競賽 優等 2024
  - 擴散模組在影像切割之應用
- 指導學生獲 台海大資訊工程學系專題競賽 亞軍 2023
  - 整合學習在醫學影像切割之應用
- 指導學生獲 台海大暑期學習實務體驗計畫 佳作 2023
- 指導學生獲 聯發科技物聯網開發競賽 入圍獎 2018
- 台海大院級研究優良教師 2014

From Notes to Knowledge: Accelerating Rare Disease Diagnosis with Explainable AI for Phenotypic-Genomic Integration

Bridging Clinical Narratives and Genomic Insights for Precision Medicine

Professor Kuan Y. Chang (張光遠)

Biomedical AI Lab, Dept. of Computer Science & Engineering,

National Taiwan Ocean University

Proposal outline

- The challenge in Rare Disease Diagnosis
- Project Objectives & Innovation
- AI Framework: Key Components
- Expected Outcomes & Validation
- Alignment with Institutional Mission

Why Rare Disease Diagnosis Needs Innovation?

Millions impacted by delayed diagnosis

Critical Gaps:

Non-structured clinical data vs. genomic data → fragmented insights

Manual analysis → prolonged diagnosis (weeks/months)

Current tools lack transparency → low clinician trust

Building a Transparent AI Diagnostic Framework

多源數據整合 | Multi-Source Data Integration

Transform free-text EHRs → structured phenotypes + genomic data

致病機轉解析 | Mechanistic Insight Generation

Model phenotype-gene associations using XAI

決策透明化 | Clinician-Centric Explainability

Traceable AI logic for trust & actionable decisions

How the XAI System Works

Input: EHR free text → NLP structuring → phenotype features

Integration: Merge with genomic data + biomedical databases (e.g., OMIM)

Analysis: XAI identifies candidate gene variants + ranks diagnoses

Output: Clinician-friendly report with evidence trails

Delivering Precision and Speed

AI Diagnostic Model

80% accuracy in prospective trials (n=50)

50% faster than manual analysis

Knowledge Database

Dynamic rare disease phenotype-genome repository

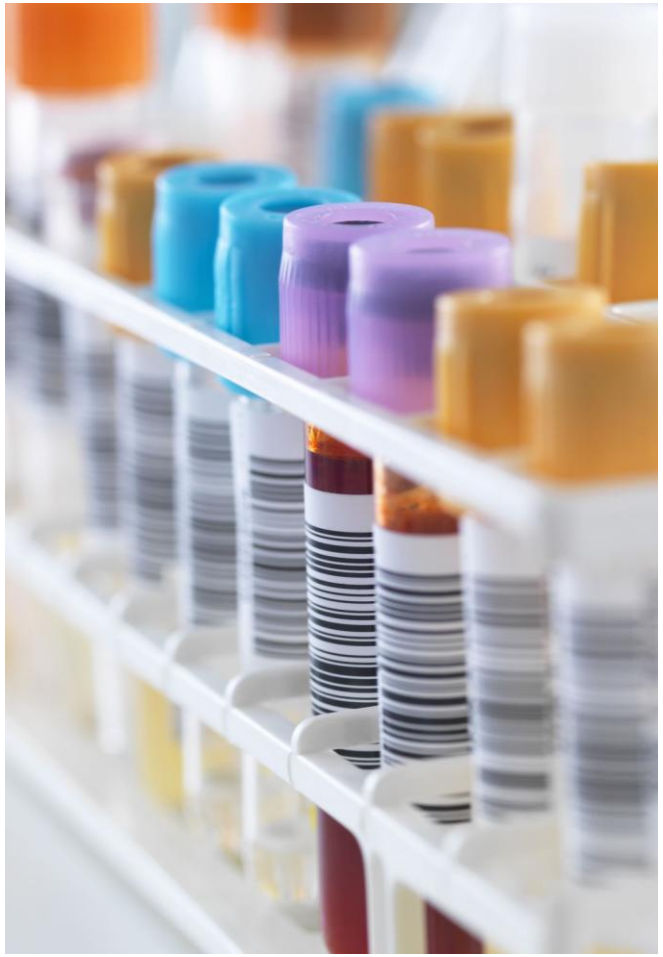
Clinical Impact

Reduced diagnostic inequality; improved patient outcomes

Advancing Precision Medicine and Equity

基隆長庚紀念醫院
Chang Gung Memorial Hospital, Keelung

國立臺灣海洋大學
NATIONAL TAIWAN OCEAN UNIVERSITY



- 精準醫學 | Precision Medicine: Bridging data silos for tailored care
- 診斷平等 | Diagnostic Equity: Democratizing access to advanced tools
- 創新可信度 | Trustworthy Innovation: Explainability fosters clinician adoption

Thank you for your attention!

Any Question?