

# 13<sup>th</sup> International Conference on Human Genetics and Genetic Diseases

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## **Nail metabopsy: An early non-invasive and affordable approach for detection of inherited metabolic disorders (IMDs) among neonates**

**Abstract:**

### **Statement of the Problem:**

Inherited metabolic disorders (IMDs) are known to represent various forms of clinical conditions with the compromised metabolic landscape of an individual that may be manifested in various stages of life starting from neonates to adolescence. Individual cases of IMD such as organic aciduria are rare, but collectively are noticeable with moderate to severe clinical manifestations. However, an approach for the early detection of IMDs among neonates is highly limited. Methodology & Theoretical Orientation: We have developed a novel methodology for the metabopsy of nail clippings by using an in-house designed VTGE system. Further, LC-HRMS was used for the detection of metabolites such as 4-hydroxyproline and organic acids to screen IMDs. Further, we have extended such observation to develop a mimetic of proline (MIPRO) against prolyl hydroxylase by using molecular docking and MD simulations. MIPRO is proposed as a potential small molecular inhibitor that prolyl hydroxylase that can alleviate the issue of high accumulation of **hydroxyproline** in tissue and free forms that leads to IMDs such as hyperhydroxyprolinemia. Findings: We have identified the high accumulation of hydroxyproline and organic acids in the nail clippings of suspected cases of IMDs among neonates. This is one of the first and novel approaches to using nail clippings for the screening of IMDs. We have designed MOPH as a potential inhibitor of prolyl hydroxylase that could serve as a proof of concept for the alleviation of IMDs. Conclusion and Significance: Taken together, the relevance of nail metabopsy are emphasized in the context of screening for IMDs such as **hyperhydroxyprolinemia** and organic aciduria. In the future, they could be explored at preclinical and clinical levels for early detection of IMDs and the development of mimetic drugs against IMDs.

### **Biography:**

Dr. Nilesh Kumar Sharma completed his Ph.D. from the Indian Institute of Technology, Roorkee in 2009 with a Health Science specialization (Free Radical Biology and Oxidative Stress). Dr. Sharma has completed post-doctoral research training for more than three years in DNA repair genes and **cancer biology** at NIEHS, NIH, USA, and Rutgers University, New Jersey Medical School, NJ, USA. Currently, a Professor (Specialization Cancer Biology and Medical Biotechnology) at DYPBBI, Dr. D. Y. Patil Vidyapeeth, Pune, India. Dr. Sharma has actively been engaged in academic work to teach subjects like Cancer Biology, Immunology, Molecular Cell Signaling, and Molecular Biology to undergraduate and postgraduate students. Dr. Sharma has been credited with more than 90 publications including in indexed National and International journals, book chapters/conference proceedings, Seven Indian patents (Published and Granting process is in progress), and Several new mimetic of metabolites are designed and submitted to PubChem.

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