



# Metabolics in Congenital Heart Problems and Phenotypes Congenital Heart Problems

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## INTRODUCTION

In the United States, there are presently 1 million people and 1 million children who have congenital heart problems, according to the Centres for Disease Control and Prevention Trusted Source. Nearly all children with heart abnormalities survive into adulthood thanks to improvements in treatments and aftercare over the previous few decades. Some people will need lifelong care for their heart defect. Nevertheless, many people manage to lead full, active lives in spite of their illness.

## DESCRIPTION

Particular metabolomic signatures have been established for a few distinct CHD subtypes. Additionally, the traits of pulmonary arterial hypertension linked to CHD have been found. This narrative review seeks to give readers an overview of the metabolomics research in the context of CHD due to the variety of the chosen studies.

Congenital Heart Disease (CHD), which affects about 8 out of every 1,000 live births, is the most prevalent birth defect. The genetic basis of CHD has been extensively studied throughout the last few decades. Undoubtedly, many CHDs are caused by genetic mutations. This is true even if distinct cardiac phenotypes can result from the same genetic background, and vice versa, diverse genetic aetiologies can result in comparable phenotypes. Additionally, epigenetics has recently drawn considerable interest. It is characterised as the study of phenotypic changes brought on by environmental changes that affect gene expression rather than changes in the genetic code.

Genetic and epigenetic connections occur in several metabolic pathways of very small molecules, some of which cross over one another. The “omics sciences” hold great promise as tools for deciphering this complex relationship. They are categorised as the areas of research that focus on compounds having relat-

ed features. They are known as “genomics,” which investigates the composition and purpose of DNA. Transcriptomics is the study of mRNA molecules; proteomics is the study of proteins, which are also a typical expression of the genome’s content.

In an effort to diagnose CHD during pregnancy, metabolomics has reportedly been widely used, according to the scientific publications that have been given. The three maternal specimens that were used were urine, blood, and amniotic fluid. Numerous unique metabolomic biomarkers have been identified, which vary depending on the metabolomics approach, the kind of maternal sample, and the trimester of pregnancy. However, it was shown that some of the effects of altered lipid metabolism and increased uric acid were similar. This is consistent with earlier research showing that babies with CHD, especially those with cyanotic heart abnormalities, have a reduced ability to use released free fatty acids in their metabolism. It seems that lipid metabolism is most frequently disrupted in this situation.

## CONCLUSION

Recent advancements in the “omics sciences” have revealed new CHD research opportunities with a variety of potential uses. A potent instrument, untargeted metabolomics has the potential to shed light on some of the more obscure characteristics that lie behind the onset of CHD. Metabolomics has the potential to distinguish between two subjects with the same genetic mutation who exhibit the cardiac defect and those who do not. This could be the next step in precision cardiology. In addition to providing potential biomarkers for the disease, identifying the affected metabolic pathways may assist in revealing the disease’s pathogenesis. However, CHD-specific metabolomics is still in its infancy. Therefore, multi-centre studies involving a large number of patients are required in order to benefit from this exciting new technology and permit its wider clinical application.

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|-------------------------|-----------------|-----------------------|-----------------------------|
| <b>Received:</b>        | 03-October-2022 | <b>Manuscript No:</b> | AASRFC-22-14989             |
| <b>Editor assigned:</b> | 05-October-2022 | <b>PreQC No:</b>      | AASRFC-22-14989 (PQ)        |
| <b>Reviewed:</b>        | 19-October-2022 | <b>QC No:</b>         | AASRFC-22-14989             |
| <b>Revised:</b>         | 24-October-2022 | <b>Manuscript No:</b> | AASRFC-22-14989 (R)         |
| <b>Published:</b>       | 31-October-2022 | <b>DOI:</b>           | 10.36648/0976-8610.13.10.95 |

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**Citation** Beets I (2022) Metabolics in Congenital Heart Problems and Phenotypes Congenital Heart Problems. Adv Appl Sci Res. 13:95.

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