



# THE METABOLIC PROFILE OF MOTHERS WITH AN ASD CHILD



This article is a review of the following research: Hollowood-Jones, K., Adams, J.B., Coleman, D.M., Ramamoorthy, S., Melnyk, S., James, S.J., Woodruff, B.K., Pollard, E.L., Snozek, C.L., Kruger, U., Chuah, J., Hahn, J. Altered metabolism of mothers of young children with Autism Spectrum Disorder: a case control study. *BMC Pediatrics*. 2020 Dec 14;20(1):557. doi: 10.1186/s12887-020-02437-7.

Metabolism is an important part of human growth and development. Metabolites are produced by cells in the body and contribute to a healthy metabolism. Metabolites include amino acids, vitamins, polyols, organic acids, nucleotides and many others.

Previous research has shown that many children with autism spectrum disorder (ASD) have low levels of certain metabolites, such as methionine, S-adenosyl methionine (SAM), cysteine, glutathione, folate, biotin, plasma ATP, plasma sulphate and plasma tryptophan.<sup>1,2,3</sup>

Researchers have wondered if similar or other abnormalities of metabolites are present in the parents of children with autism. Some studies have shown differences in specific metabolites in mothers, such as folate and sulphur.<sup>4,5</sup> However, further research is needed to investigate metabolites as a whole. Researchers from Rensselaer Polytechnic Institute, Arizona State University and Mayo Clinic collaborated to investigate the entire metabolic profile of mothers of children with autism.

## Study

- The research team analyzed blood samples from 30 mothers of children with autism (ASD-M) and 29 mothers of typically developing children (TD-M).
- Samples were taken when the children were between the ages of two and five years old.
- The research team measured a total of 622 metabolites in blood samples, including levels of vitamin B12, folate, methylmalonic acid, homocysteine, isoprostane, vitamin D, vitamin E, hCG and MTHFR variants.
- Researchers took special note of the metabolic analysis of the folate-mediated one-carbon metabolism (FOCM) and the transsulfuration pathway (TS) since previous research has shown significant results in these areas.

## Results

Overall, the analysis showed that mothers of children with ASD (ASD-M) and mothers of typically developing children (TD-M) have significantly different metabolic profiles. The research group identified the top 50 metabolites that show the most significant differences between TD-M and ASD-M. In almost every case, the ASD-M had lower levels of the metabolite than the TD-M.

- The research group found that ASD-M had significantly lower levels of vitamin B12, and carnitine-conjugated metabolites than the TD-M.
- The results did not specifically show lower levels of folate in the ASD-M, although other metabolites, closely linked to folate, were significantly lower.
- The ASD-M had especially low levels of 4-vinylphenol sulfate, NAD<sup>+</sup>, and 3 glycine containing metabolites.
- The researchers grouped these differences into five core metabolite groups, where the following metabolites are representative of each group: Glu-Cys, Histidylglutamate, Cinnamoylglycine, Proline, and Adrenoylcarnitine.

To confirm the results, the researchers took the data from the five core groups of metabolites and worked backwards to see if they could predict if the sample came from a mother belonging to the ASD-M or TD-M group. The analysis differentiated the two groups (ASD-M and TD-M) with 97 percent accuracy.

## Interpretation of the Research

This research shows that mothers with a child on the spectrum have a significantly different metabolic profile than mothers with a typically developing child. Some of the biggest differences were associated with the carnitine metabolism. Some carnitines are produced by the body, but the primary sources of carnitine are the consumption of beef and pork. The fact that the ASD-M and the TD-M did not report any differences in beef/pork consumption means that there must be a difference in how the two groups process carnitine. Carnitine is known to improve mitochondrial function and increase cellular energy. It is worth noting that some studies have suggested that a carnitine supplement might benefit children with autism.<sup>6,7</sup> Could this also be true for mothers with a child on the spectrum?

Significant differences were also noted with Vitamin B12, which helps in the formation of red blood cells and in the maintenance of the central nervous system. Could mothers with a child with autism benefit from taking a vitamin B12 supplement?

This study did not look at the father's metabolic profile, which can be affected by his genetics, diet and environmental exposure. The question remains as to what role his genetic/metabolic profile plays in the picture of the autism family.

Another area of research is determining if these differences are seen in mothers during pregnancy. If metabolic differences are seen during pregnancy, there is potential to develop a blood test that will screen mothers who are at a higher risk of having a child with ASD. The research team is currently working on this project.

It's important to note that further studies with larger sample sizes are needed to validate these findings. However, the results are promising and have the potential to increase our understanding of autism. We look forward to reading the future findings of this team's research.

Written by Autism Advocate Parenting Magazine

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