

Genetics

Newborn screening for MCAD deficiency

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Medium chain acyl-CoA dehydrogenase (MCAD) deficiency is an autosomal recessive fatty acid oxidation disorder, with an incidence rate of up to 1 in 12000. Early diagnosis and treatment might prevent sudden death or long-term disability.

Bottom line. Parents-to-be should be informed, preferably in the prenatal period, of newborn screening for MCAD deficiency and other disorders. The panel of disorders screened for during that time varies by province—physicians should familiarize themselves with their local newborn screening programs.

The complete *Gene Messenger—Newborn Screening for MCAD Deficiency* by the GenetiKit research team is available on **CFPlus**.^{*} Past Gene Messenger articles can be accessed on-line at www.cfp.ca. On the home page, click on **Collections** in the left-hand menu, then click on **Genetics**.

Competing interests

None declared

The **GenetiKit** research team, a group of family physicians, genetic counselors and geneticists, designed the Gene Messenger series to provide practical information to help family physicians and their patients make informed choices about rapidly emerging genetic discoveries. The series is a collection of up-to-date, definitive, short reviews on genetics topics that have made headlines, and offers recommendations regarding referral for genetic services or testing.

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GENE MESSENGER

For more information on genetics topics,
see www.mtsinai.on.ca/FamMedGen/



^{*}The Gene Messenger on newborn screening for MCAD deficiency is available at www.cfp.ca. Go to the full text of this article on-line, then click on **CFPlus** in the menu at the top right-hand side of the page.