

POLG1 Mutation in Pregnancy

Date of Search: 10/08/2016

Sources Searched: Medline, Embase, Google Scholar

Search History:

1. EMBASE; (polg1 adj2 mutat*).ti,ab; 144 results.
2. EMBASE; ("Mitochondrial DNA polymerase*" adj2 mutat*).ti,ab; 25 results.
3. EMBASE; exp DNA DIRECTED DNA POLYMERASE GAMMA/; 1713 results.
4. EMBASE; exp GENE MUTATION/; 508747 results.
5. EMBASE; 3 AND 4; 474 results.
6. EMBASE; 1 OR 2 OR 5; 594 results.
7. EMBASE; preg*.ti,ab; 520621 results.
8. EMBASE; exp PREGNANCY/; 624507 results.
9. EMBASE; 7 OR 8; 813663 results.
10. EMBASE; 6 AND 9; 4 results.
11. EMBASE; *INTRACTABLE EPILEPSY/; 2734 results.
12. EMBASE; 9 AND 11; 18 results.
13. EMBASE; exp PREGNANCY COMPLICATION/; 116600 results.
14. EMBASE; 11 AND 13; 1 results.
15. EMBASE; exp INTRACTABLE EPILEPSY/; 6503 results.
16. EMBASE; 13 AND 15; 11 results.
17. EMBASE; exp DISORDERS OF MITOCHONDRIAL FUNCTIONS/; 29201 results.
18. EMBASE; 13 AND 17; 117 results.
19. EMBASE; exp EPILEPSY/; 193207 results.
20. EMBASE; 18 AND 19; 4 results.
21. EMBASE; exp ALPERS DISEASE/; 275 results.
22. EMBASE; 9 AND 21; 5 results.
23. EMBASE; exp GENE MUTATION/ AND exp MITOCHONDRIAL DNA/; 8915 results.
24. EMBASE; 13 AND 23; 22 results.
25. EMBASE; 3 AND 13; 0 results.
26. EMBASE; 3 AND 9; 18 results.
27. EMBASE; exp EPILEPSY/ AND exp DISORDERS OF MITOCHONDRIAL FUNCTIONS/; 2154 results.
28. EMBASE; 13 AND 27; 4 results.
29. EMBASE; 9 AND 27; 30 results.
30. EMBASE; 6 AND 15; 15 results.
31. EMBASE; exp PREECLAMPSIA/; 43901 results.
32. EMBASE; 6 AND 31; 0 results.
33. EMBASE; 17 AND 31; 48 results.

34. EMBASE; polg.ti,ab; 636 results.
35. EMBASE; 31 AND 34; 0 results.
36. EMBASE; 9 AND 34; 10 results.
37. EMBASE; 13 AND 34; 0 results.
38. Medline; exp MITOCHONDRIAL DISEASES/; 12742 results.
39. Medline; (polg1 adj2 mutat*).ti,ab; 96 results.
40. Medline; ("Mitochondrial DNA polymerase*" adj2 mutat*).ti,ab; 35 results.
41. Medline; 38 OR 39 OR 40; 12804 results.
42. Medline; preg*.ti,ab; 379622 results.
43. Medline; exp PREGNANCY/; 790627 results.
44. Medline; 42 OR 43; 866036 results.
45. Medline; 41 AND 44; 284 results.
46. Medline; 39 AND 44; 0 results.
47. Medline; 40 AND 44; 0 results.
48. Medline; epileps*.ti,ab; 81845 results.
49. Medline; exp DRUG RESISTANT EPILEPSY/; 192 results.
50. Medline; 48 OR 49; 81867 results.
51. Medline; 45 AND 50; 4 results.
52. Medline; 44 AND 49; 1 results.
53. Medline; exp PREGNANCY COMPLICATIONS/; 375293 results.
54. Medline; 41 AND 53; 135 results.
55. Medline; 50 AND 54; 2 results.
56. Medline; preeclamps*.ti,ab; 22437 results.
57. Medline; exp PRE-ECLAMPSIA/; 26007 results.
58. Medline; 56 OR 57; 34201 results.
59. Medline; 41 AND 58; 15 results.
60. Medline; exp MELAS SYNDROME/; 1089 results.
61. Medline; 58 AND 60; 1 results.
62. Medline; 53 AND 60; 13 results.
63. EMBASE; exp MELAS SYNDROME/; 2119 results.
64. EMBASE; 9 AND 63; 25 results.
65. EMBASE; 17 AND 31; 48 results.
66. EMBASE; 27 AND 31; 2 results.
67. EMBASE; exp SEIZURE, EPILEPSY AND CONVULSION/; 296555 results.
68. EMBASE; 9 AND 17 AND 67; 56 results.
69. EMBASE; exp LABOR/ OR exp LABOR COMPLICATION/; 182740 results.
70. EMBASE; 27 AND 69; 22 results.
71. EMBASE; 6 AND 69; 0 results.
72. EMBASE; 17 AND 69; 150 results.
73. EMBASE; epilep*.ti,ab; 153678 results.
74. EMBASE; 72 AND 73; 7 results.
75. EMBASE; *LABOR/ OR *LABOR COMPLICATION/; 23136 results.
76. EMBASE; 17 AND 75; 1 results.
77. EMBASE; exp MITOCHONDRIAL MYOPATHY/; 2495 results.
78. EMBASE; 13 AND 77; 6 results.
79. EMBASE; 9 AND 77; 31 results.
80. EMBASE; exp CEREBROVASCULAR ACCIDENT/; 129758 results.

81. EMBASE; 9 AND 17 AND 80; 12 results.
82. EMBASE; 9 AND 23 AND 80; 0 results.
83. Medline; stroke.ti,ab; 171639 results.
84. Medline; exp STROKE/; 101457 results.
85. Medline; 83 OR 84; 204080 results.
86. Medline; 45 AND 85; 12 results.
87. Medline; melas.ti,ab; 1593 results.
88. Medline; 44 AND 87; 28 results.
89. Medline; exp MITOCHONDRIAL ENCEPHALOMYOPATHIES/; 2113 results.
90. Medline; 44 AND 89; 30 results.
91. Medline; exp OBSTETRIC LABOR COMPLICATIONS/; 58627 results.
92. Medline; 41 AND 91; 6 results.
93. Medline; 38 AND 91; 6 results.
94. Medline; "alper's disease".ti,ab; 3 results.
95. Medline; polg.ti,ab; 384 results.
96. Medline; 44 AND 95; 5 results.
100. EMBASE; *DISORDERS OF MITOCHONDRIAL FUNCTIONS/; 5589 results.
101. EMBASE; 9 AND 100; 77 results.

Other Useful Sources:

The Newcastle Mitochondrial Disease Guidelines: Pregnancy

http://www.mitochondrialncg.nhs.uk/documents/Pregnancy_Guidelines_%202011.pdf

Title: Mitochondrial disease in pregnancy: a systematic review

Citation: Obstetric Medicine. 2011 4 [3] pp. 90-94

Author(s): Say RE, Whittaker RG, Turnbull HE, McFarland R, Taylor RW, Turnbull DM

Title: A systematic review of mitochondrial disease in pregnancy

Citation: Archives of Disease in Childhood: Fetal and Neonatal Edition, June 2010, vol./is. 95/(Fa54), 1359-2998 (June 2010)

Author(s): Say R.E., Whittaker R.G., Turnbull H.E., McFarland R., Taylor R.W., Turnbull D.M.

Language: English

Abstract: Background Mitochondrial diseases are heterogeneous in clinical presentation and genotype. The incidence of known pathogenic mitochondrial DNA mutations in the general population is 1 in 500. Little is known about the implications of pregnancy for women with mitochondrial disease. Methods The authors undertook a systematic review of the literature on mitochondrial disease in pregnancy. Results Ten case reports were identified. The most common complications were threatened preterm labour (five women) and pre-eclampsia (four women). Two women experienced magnesium sulphate toxicity. Pregnancy had a varied effect on mitochondrial disease with some women being asymptomatic; others

developing mild symptoms such as exercise intolerance or muscle weakness which resolved postnatally; and others developed more serious, persistent symptoms such as symptomatic Wolff- Parkinson-White syndrome, persistent paraesthesia and focal segmental glomerulosclerosis. Discussion Women with mitochondrial disease appear to be at increased risk of complications during pregnancy and labour but further prospective cohort studies are needed.

Publication Type: Journal: Conference Abstract

Source: EMBASE

Full Text:

Available from *Highwire Press* in [Fetal and Neonatal](#)

Title: Pregnancy and delivery under the MELAS mutation

Citation: Mitochondrion, March 2016, vol./is. 27/(39), 1567-7249;1872-8278 (March 01, 2016)

Author(s): Finsterer J., Frank M.

Language: English

Publication Type: Journal: Letter

Source: EMBASE

Title: Obstetric complications in carriers of the m.3243A>G mutation, a retrospective cohort study on maternal and fetal outcome

Citation: Mitochondrion, November 2015, vol./is. 25/(98-103), 1567-7249;1872-8278 (November 01, 2015)

Author(s): de Laat P., Fleuren L.H.J., Bekker M.N., Smeitink J.A.M., Janssen M.C.H.

Language: English

Abstract: Introduction: The mitochondrial DNA m.3243A. > G mutation is the most prevalent mutation causing mitochondrial disease in adult patients. Aside from some case reports, there are no studies on obstetric complications in a cohort of m.3243A > G carriers. We aimed to identify the prevalence of obstetric complications in a cohort of women carrying the m.3243A > G mutation. Methods: All female carriers of the m.3243A > G mutation known from our previous national inventory were sent a questionnaire regarding their obstetric history. Data were compared to national references. Data from the national inventory, including NMDAS (disease severity) scores and heteroplasmy levels in urinary

epithelial cells (UEC) were used to stratify women. Results: Sixty women participated, the mean age was 47 years (range 20-70), mean NMDAS was 14.6 (range 0-46), and mean heteroplasmy percentage in UEC was 19.9% (range 5-85%). Ninety-eight pregnancies in 46 women were reported. Twenty-three (25.3%) had a premature delivery and five of them (5.5%) had a gestation of < 32 weeks and eleven of the women (12%) suffered from preeclampsia. No different heteroplasmy level was found in the women with preeclampsia. Nine pregnancies (11%) were complicated by gestational diabetes. Discussion: Obstetric complications occur frequently in carriers of the m.3243A > G mutation. Proper guidance during pregnancies and early detection of possible obstetric complications are needed. As techniques to prevent transmission of mitochondrial mutations are studied it is important to know the possible complications patients may experience from the ensuing pregnancy.

Publication Type: Journal: Article

Source: EMBASE

Title: The mitochondrion, its genome and their contribution to well-being and disease

Citation: Molecular Human Reproduction, June 2014, vol./is. 21/1(1-2), 1360-9947;1460-2407 (19 Jun 2014)

Author(s): St. John J.C.

Language: English

Publication Type: Journal: Editorial

Source: EMBASE

Full Text:

Available from *Highwire Press* in [Molecular Human Reproduction](#)

Available from *Oxford University Press* in [MHR: Basic Science of Reproductive Medicine](#);

Note: ; Collection notes: To access please select Login with Athens and search and select NHS England as your institution before entering your NHS OpenAthens account details.

Title: Maternal blood mitochondrial DNA copy number and placental abruption risk: Results from a preliminary study

Citation: International Journal of Molecular Epidemiology and Genetics, 2013, vol./is. 4/2(120-127), 1948-1756 (2013)

Author(s): Williams M.A., Sanchez S.E., Ananth C.V., Hevner K., Qiu C., Enquobahrie D.A.

Language: English

Abstract: Oxidative stress and impaired placental function - pathways implicated in the pathogenesis of placental abruption - have their origins extending to mitochondrial dysfunction. To the best of our knowledge, there are no published reports of associations of placental abruption with circulating mitochondrial DNA (mtDNA) copy number - a novel biomarker of systemic mitochondrial dysfunction. This pilot case-control study was comprised of 233 placental abruption cases and 238 non-abruption controls. Real-time quantitative polymerase chain reaction (PCR) was used to assess the relative copy number of mtDNA in maternal whole blood samples collected at delivery. Logistic regression procedures were used to estimate adjusted odds ratios (OR) and 95% confidence intervals (CI). There was some evidence of an increased odds of placental abruption with the highest quartile of mtDNA copy number (P for trend = 0.09) after controlling for confounders. The odds of placental abruption was elevated among women with higher mtDNA copy number (>336.9) as compared with those with lower values (<336.9) (adjusted OR = 1.60; 95% CI 1.04-2.46). Women diagnosed with preeclampsia and with elevated mtDNA copy number had a dramatically increased odds of placental abruption as compared with normotensive women without elevated mtDNA copy number (adjusted OR = 6.66; 95% CI 2.58-17.16). Maternal mitochondrial dysfunction appears to be associated with placental abruption in the presence of preeclampsia. Replication in other studies, particularly prospective cohort studies and those that allow for tissue specific assessment of mitochondrial dysfunction (e.g., the placenta) are needed to further understand cellular and genomic biomarkers of normal and abnormal placental function.

Publication Type: Journal: Article

Source: EMBASE

Full Text:

Available from *National Library of Medicine* in [International Journal of Molecular Epidemiology and Genetics](#)

Title: Mitochondrial disease: Fetal and maternal presentations and issues in pregnancy

Citation: Molecular Genetics and Metabolism, March 2012, vol./is. 105/3(282), 1096-7192 (March 2012)

Author(s): Feigenbaum A.

Language: English

Abstract: Mitochondrial diseases, clinical illness resulting from defects in cellular energy metabolism, are common. Prevalence has been reported at ~1:5-10,000. Knowledge about these diseases is expanding rapidly and all health care practitioners should be aware of the fundamental principles and presentations. I will discuss some of these as an introduction to the rest of the day's talks about mitochondrial diseases. For the purposes of this presentation I will refer to primary conditions affecting pyruvate metabolism and the respiratory chain, both defects of the mtDNA and nuclear-encoded proteins as well as those involved in the transcription, replication and signaling pathways. I will use case examples

and literature review to illustrate the challenges facing us regarding the genetics and inheritance patterns, fetal presentations and maternal issues during pregnancy and labour.

Publication Type: Journal: Conference Abstract

Source: EMBASE

Title: Anaesthesia in patient with the melas syndrome for cesarean section [Croatian]
Anestezija za carski rez kod rodilje s melas sindromom

Citation: Gynaecologia et Perinatologia, 2012, vol./is. 22/1(22-24), 1330-0091 (2012)

Author(s): Marijic V., Matas M., Marohnic R., Mihaljevic S., Mihaljevic L.

Language: Croatian

Abstract: We present a case of a parturient in 34th week of gestational age with the MELAS syndrome whose pregnancy ended with cesarean section because of threatening fetal asphyxiation. The MELAS syndrome is a rare mitochondriopathy with multigrain presentation and diverse clinical picture, which is most often characterized with myopathy, encephalopathy, lactic acidosis, convulsions and stroke like episodes. Our patient was preoperatively hemodynamically stable, and despite using sevoflurane and succinylcholine there were no signs of malignant hyperthermia as no other complications otherwise possible during general anesthesia.

Publication Type: Journal: Article

Source: EMBASE

Full Text:

Available from *Free Access Content* in [Gynaecologia et Perinatologia](#)

Title: Anaesthetic considerations for caesarean section in a patient with mitochondrial myopathy (complex I and IV deficiency) with respiratory weakness: A case report

Citation: International Journal of Obstetric Anesthesia, May 2010, vol./is. 19/(S30), 0959-289X (May 2010)

Author(s): Braham D.L., Bell R.

Language: English

Abstract: Introduction: Mitochondrial myopathies are a rare heterogeneous group of disorders caused by abnormalities of mitochondrial DNA resulting in defects of the respiratory chain. Although multiple organ systems may be affected, they predominantly

affect skeletal muscle. Three groups of mitochondrial abnormalities are described: the respiratory chain deficiencies (complexes I-IV), mitochondrial DNA mutations and mitochondrial deletions. 1 Case Report: A 28-year old patient with a mitochondrial respiratory chain defect affecting complexes I (NADH oxidase - absence of activity) and IV (cytochrome oxidase-reduced activity), diagnosed at 10 years of age was referred to the multi-disciplinary high-risk obstetric team. Her main problems were skeletal myopathy with poor mobility and respiratory weakness, elevated blood lactate, cataracts, pigment retinopathy and short stature. She had documented nocturnal hypoventilation treated with CPAP (6cm H₂O) for the last 7 years. She was monitored during pregnancy with lung function tests, sleep studies and serial fetal growth scans but showed no evidence of deterioration. Elective caesarean section (CS) was undertaken at 37 weeks of gestation. Full standard anaesthetic monitoring was instituted. Supplemental face mask oxygen was given to increase the baseline oxygen saturation from 93% to 97%. We performed combined spinal-epidural anaesthesia with 12 mg bupivacaine and 400 meg diamorphine intrathecally. A further 50 mg bupivacaine was given epidurally to achieve a block to T4. A phenylephrine infusion was titrated to maintain baseline blood pressure until delivery. There were no episodes of dyspnoea or desaturation therefore intraoperative CPAP was not required. Post-natal progress was unremarkable. Discussion: Patients with mitochondrial myopathies can exhibit a variety of different clinical syndromes. Any organ system can be involved necessitating individualised management. The main issues for our patient were respiratory weakness and lactic acidosis. CS was the delivery mode of choice in order to avoid the increased metabolic demand of labour which might precipitate potentially life threatening lactic acidosis or respiratory compromise.² Regional anaesthesia reduces metabolic stress and avoids the problems of general anaesthesia in these patients which may include potential drug sensitivity (induction agents and nondepolarizing muscle relaxants), malignant hyperthermia, hyperkalaemia (suxamethonium use) 3 and post-operative respiratory depression.

Publication Type: Journal: Conference Abstract

Source: EMBASE

Title: The risk of complications in pregnancy and labour for women with mitochondrial disease

Citation: Journal of Neurology, Neurosurgery and Psychiatry, November 2009, vol./is. 80/11(no pagination), 0022-3050 (November 2009)

Author(s): Turnbull H., Whittaker R.G., Phillips A., Poulton J., McFarland R.

Language: English

Abstract: In patients with mitochondrial disease it might be anticipated that the increased energy demand during pregnancy and particularly at the onset of labour, may lead to serious complications. We devised a comprehensive questionnaire to assess the risk of these complications. Questionnaires were posted to 63 female patients with genetically

confirmed mitochondrial disease (m.3243A.G or large-scale Single Deletion (SD)) identified from a cohort of patients attending a specialist mitochondrial disease centre. Questionnaires were returned by 31 (49%) individuals and included details on 80 pregnancies. Pregnancy induced hypertension was more frequent in the m.3243A. G group (31.4%) compared with the SD group (8.9%) and published controls (6-17.4%). Diabetes mellitus was a complication encountered in 5.7% of m.3243A.G patient pregnancies, none of the SD patients and 0.41% of published control data. The frequency of instrumental delivery and caesarean section were comparable to current UK estimates, but the number of premature births ((36 weeks) was greater than expected. Our data indicate that specific complications of pregnancy occur more frequently in patients with the m.3243A.G mutation, but that this finding cannot be generalised to all forms of mitochondrial disease. We recommend increased vigilance in the obstetric management of m.3243A.G patients.

Publication Type: Journal: Conference Abstract

Source: EMBASE

Full Text:

Available from *HighWire Press* in [Journal of neurology, neurosurgery, and psychiatry](#)

Available from *ProQuest* in [Journal of Neurology, Neurosurgery and Psychiatry](#)

Title: Successful pregnancy and cesarean delivery via noninvasive ventilation in mitochondrial myopathy

Citation: Journal of Perinatology, 2009, vol./is. 29/2(166-167), 0743-8346;1476-5543 (2009)

Author(s): Yuan N., El-Sayed Y.Y., Ruoss S.J., Riley E., Enns G.M., Robinson T.E.

Language: English

Abstract: We report a case study of a 22-year-old woman with mitochondrial thymidine kinase 2 deficiency and chronic respiratory failure due to severe neuromuscular weakness requiring noninvasive positive pressure ventilation (NIPPV) since 12 years of age. During pregnancy and cesarean delivery, she was successfully supported with NIPPV. A multidisciplinary team approach should be used in pregnant patients with these disorders with specific attention to management of pulmonary complications, selection of route of delivery, anesthesia, and analgesia.

Publication Type: Journal: Article

Source: EMBASE

Full Text:

Available from *Nature Publishing Group* in [Journal of Perinatology](#)

Available from *ProQuest* in [Journal of Perinatology](#)

Title: Anesthetic management of an obstetric patient with MELAS syndrome: case report and literature review.

Citation: International journal of obstetric anesthesia, Oct 2008, vol. 17, no. 4, p. 370-373, 1532-3374 (October 2008)

Author(s): Maurtua, M, Torres, A, Ibarra, V, DeBoer, G, Dolak, J

Abstract: Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS syndrome) is a mitochondrial disorder associated with neurologic, cardiac, neuromuscular, hepatic, metabolic and gastrointestinal dysfunction and potential anesthetic and obstetric complications. The case of a parturient with MELAS syndrome requiring labor analgesia is presented. A Medline literature search limited to the English language was undertaken to review cases of MELAS syndrome. Based on our experience and literature review, parturients with MELAS syndrome appear to benefit from neuraxial analgesia and anesthesia, which blunt excessive oxygen consumption and acidosis.

Source: Medline

Title: Pre-eclampsia and magnesium toxicity with therapeutic plasma level in a woman with m.3243A > G melas mutation

Citation: Journal of Obstetrics and Gynaecology, April 2008, vol./is. 28/3(349), 0144-3615;1364-6893 (April 2008)

Author(s): Moriarty K.T., McFarland R., Whittaker R., Burch J., Turnbull H.E., Taylor R.W., Turnbull D.M.

Language: English

Publication Type: Journal: Article

Source: EMBASE

Full Text:

Available from *Taylor & Francis* in [Journal of Obstetrics and Gynaecology](#)

Title: Two full-term pregnancies in a patient with mitochondrial myopathy and chronic ventilatory insufficiency

Citation: Respiration, November 2005, vol./is. 72/6(654-656), 0025-7931 (November/December 2005)

Author(s): Diaz-Lobato S., Gomez Mendieta M.A., Moreno Garcia M.S., Mayoralas-Alises S., Arpa Gutierrez F.J.

Language: English

Abstract: Mitochondrial myopathies are a group of diseases characterized by metabolic defects at the mitochondrial respiratory chain level which result in impaired oxidative phosphorylation and ATP synthesis. As with other neuromuscular diseases, respiratory muscles can be affected and ventilatory failure may occur. There have been isolated case reports of pregnant patients with ventilatory failure due to neuromuscular diseases such as polio and spinal muscular atrophy. We describe the case of a 34-year-old patient with mitochondrial myopathy and ventilatory failure requiring non-invasive ventilation who carried two pregnancies to term with no complications. We have not found a similar case in the literature. Copyright © 2005 S. Karger AG.

Publication Type: Journal: Article

Source: EMBASE

Full Text:

Available from *ProQuest* in [Respiration](#)

Title: Mitochondrial disease and pregnancy [Spanish] Enfermedades mitocondriales y gestacion

Citation: Clinica e Investigacion en Ginecologia y Obstetricia, March 2005, vol./is. 32/2(61-77), 0210-573X (March/April 2005)

Author(s): Tejerizo-Garcia A., Hernandez-Hernandez L., Henriquez A., Gonzalez-Rodriguez S.P., Ruiz M.A., Alcantara R., Martinez Del Val M.P., Lanchares J.L., Tejerizo-Lopez L.C.

Language: Spanish

Abstract: We report the case of a primigravid woman with a mitochondrial disorder affecting the energy-generating pathway of oxidative phosphorylation discovered 3 years after the appearance of progressive proximal tubular renal insufficiency, muscular weakness, bilateral optical neuropathy and diffuse leucoencephalopathy detected on magnetic resonance imaging of the brain. Her diagnosis was made on the basis of abnormal mitochondria on a muscle biopsy and of spectrophotometric deficiencies of the complexes I, II, III and IV of the respiratory chain. The search for specific genetic mutation was negative. Her pregnancy was complicated by severe preeclampsia, gestational diabetes requiring insulin, and severe renal failure, necessitating premature delivery by cesarean section at 30 weeks gestation. The evolution of the 1070g female neonate was uneventful, and at two years of age she shows no signs of mitochondrial disease. However, the mother's postpartum was complicated by seizures and terminal renal failure, for which she is currently having dialysis whilst waiting for a renal transplant in the near future.

Publication Type: Journal: Review

Source: EMBASE

Full Text:

Available from *Free Access Content* in [Clínica e Investigación en Ginecología y Obstetricia](#)

Title: [Pregnancy in a patient with mitochondrial disease].

Citation: Journal de gynécologie, obstétrique et biologie de la reproduction, Apr 2004, vol. 33, no. 2, p. 131-139, 0368-2315 (April 2004)

Author(s): Racine, A-C, Blanchot, G, Le Vaillant, C, Boog, G

Abstract: We report a case of a pregnant woman with a mitochondrial disorder affecting the energy-generating pathway of oxidative phosphorylation which was suggested when the patient presented the progressive clinical phenotype of a proximal tubular renal insufficiency, a muscular weakness of extremities, a bilateral optic neuropathy and a brain magnetic resonance imaging suggesting diffuse leucoencephalopathy. Her diagnosis was made on the basis of abnormal mitochondria on a muscle biopsy and of spectrophotometric deficiencies of the complexes I, II+III and IV of the respiratory chain. No specific molecular mutation could be detected. Her pregnancy was complicated by a severe preeclampsia, an insulin requiring gestational diabetes and a worrying renal failure which precipitated the premature delivery by cesarean section at 30 weeks gestation. The clinical course of the female neonate weighing 1030 grams was uneventful. At two Years of age she showed no sign of mitochondrial disease. But the postpartum course of the mother was complicated by seizures and a terminal renal failure leading presently to dialysis, but requiring a kidney transplantation in the near future.

Source: Medline

Title: Postpartum hemorrhage and emergency hysterectomy in a patient with mitochondrial myopathy: a case report.

Citation: Archives of gynecology and obstetrics, Feb 2003, vol. 267, no. 4, p. 247-249, 0932-0067 (February 2003)

Author(s): Dessole, Salvatore, Capobianco, Giampiero, Ambrosini, Guido, Battista Nardelli, Giovanni

Abstract: Mitochondrial myopathies are a rare biochemical group of disorders of the mitochondrial respiratory chain. We report the first case in the literature of a pregnant woman with mitochondrial myopathy who, after cesarean section, had a severe and massive postpartum hemorrhage that required emergency supracervical hysterectomy. We discuss the case and review the literature.

Source: Medline

Full Text:

Available from *Springer Link Journals* in [Archives of Gynecology and Obstetrics](#)

Title: Pregnancy with mitochondrial encephalopathy, lactic acidosis, and strokelike episodes syndrome.

Citation: *Obstetrics and gynecology*, May 1999, vol. 93, no. 5 Pt 2, p. 853., 0029-7844 (May 1999)

Author(s): Kovilam, O P, Cahill, W, Siddiqi, T A

Source: Medline

Full Text:

Available from *Obstetrics and Gynecology* in [Patricia Bowen Library and Knowledge Service West Middlesex university Hospital](#)

Available from *Ovid* in [Obstetrics and Gynecology](#)

Title: Mitochondrial myopathy in a primigravid pregnancy

Citation: *British Journal of Obstetrics and Gynaecology*, 1999, vol./is. 106/8(871-873), 0306-5456 (1999)

Author(s): Blake L.L., Shaw R.W.

Language: English

Publication Type: Journal: Article

Source: EMBASE

Full Text:

Available from *British Journal of Obstetrics and Gynaecology* in [Patricia Bowen Library and Knowledge Service West Middlesex university Hospital](#)

Title: Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes with deterioration during pregnancy.

Citation: *Internal medicine (Tokyo, Japan)*, Sep 1998, vol. 37, no. 9, p. 780-783, 0918-2918 (September 1998)

Author(s): Yanagawa, T, Sakaguchi, H, Nakao, T, Sasaki, H, Matsumoto, G, Sanke, T, Nanjo, K

Abstract: We report a 31-year-old woman who developed myopathy and neuropathy during pregnancy. She was diagnosed as having mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS). A T-to-C transition mutation at nucleotide position 3271 was detected in the mitochondrial gene. Her symptoms subsided spontaneously and she delivered a male infant at 38 weeks of gestation. Prior reports on

mitochondrial diseases with pregnancy are very rare, probably because of the early onset of the disease. The metabolic changes during pregnancy increase the stress on the mitochondrial function, particularly in patients with impaired mitochondrial function. Therefore pregnancy can aggravate mitochondrial diseases.

Source: Medline

Title: Pregnancy and delivery complicated by mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes.

Citation: *Obstetrics and gynecology*, May 1998, vol. 91, no. 5 Pt 2, p. 865., 0029-7844 (May 1998)

Author(s): Kokawa, N, Ishii, Y, Yamoto, M, Nakano, R

Source: Medline

Full Text:

Available from *Obstetrics and Gynecology* in [Patricia Bowen Library and Knowledge Service West Middlesex university Hospital](#)

Available from *Ovid* in [Obstetrics and Gynecology](#)

Title: Anaesthetic management of labour and delivery in the parturient with mitochondrial myopathy

Citation: *Canadian Journal of Anaesthesia*, April 1996, vol./is. 43/4(403-407), 0832-610X (April 1996)

Author(s): Rosaeg O.P., Morrison S., MacLeod J.P.

Language: English

Abstract: Purpose: We describe the anaesthetic management for Caesarean section in a parturient with a defect in complex III of the respiratory chain who had increased lactate concentrations at rest and with exercise. Clinical features: We administered effective epidural anaesthesia with lidocaine for Caesarean delivery. The serum lactate concentration was less than the preoperative value both during and after surgery. Shivering during the perioperative period was avoided by administering warm iv fluids, warm local anaesthetic solution and epidural meperidine. Pain relief after surgery was provided with iv PCA morphine augmented by local infiltration with bupivacaine to fascia and skin edges and epidural injection with meperidine. Conclusion: Mitochondrial myopathies are an uncommon group of disorders in which mitochondrial dysfunction leads to clinical disease of muscle and sometimes of other organs with high energy requirements. The management of labour and delivery in women with mitochondrial myopathies should be individualized according to severity of disease and formulated by consultation between attending physicians and the anaesthetist. Epidural analgesia reduces stress at work associated with

labour and reduces oxygen demand during labour However, parturients with defects of the respiratory chain with documented increased lactate concentrations at rest and with exercise are best managed with elective Caesarean delivery with regional anaesthesia to prevent life-threatening lactic acidosis during labour. The association between malignant hyperthermia and these disorders have not been proved, but it appears prudent to consider these women as MH susceptible until definitive data regarding the possible relationship are available.

Publication Type: Journal: Article

Source: EMBASE

Full Text:

Available from *Springer Link Journals* in [Canadian Journal of Anesthesia/Journal canadien d'anesthésie](#)

Available from *Free Access Content* in [Canadian Journal of Anesthesia](#)

Title: Mitochondrial myopathy and preeclampsia associated with pregnancy

Citation: American Journal of Obstetrics and Gynecology, 1990, vol./is. 162/1(146-147), 0002-9378 (1990)

Author(s): Berkowitz K., Monteagudo A., Marks F., Jackson U., Baxi L.

Language: English

Abstract: Mitochondrial myopathy is characterized by weakness, exercise intolerance, and acidosis. Pregnancy has been reported to accelerate the disease process. This report discusses pregnancy and management of labor complication by mitochondrial myopathy and the therapeutic dilemmas that arise when preeclampsia is diagnosed.

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