

## 신생아기에 진단된 미토콘드리아 호흡 사슬 결함 1례

연세대학교 의과대학 소아과학교실<sup>1</sup> · 임상유전학과<sup>2</sup>

최경민<sup>1</sup> · 권해식<sup>1</sup> · 이동우<sup>1</sup> · 남궁란<sup>1</sup> · 박민수<sup>1</sup> · 이철<sup>1</sup> · 이영목<sup>2</sup> · 이진성<sup>2</sup>

### A Case of Mitochondrial Respiratory Chain Defect Diagnosed in the Neonatal Period

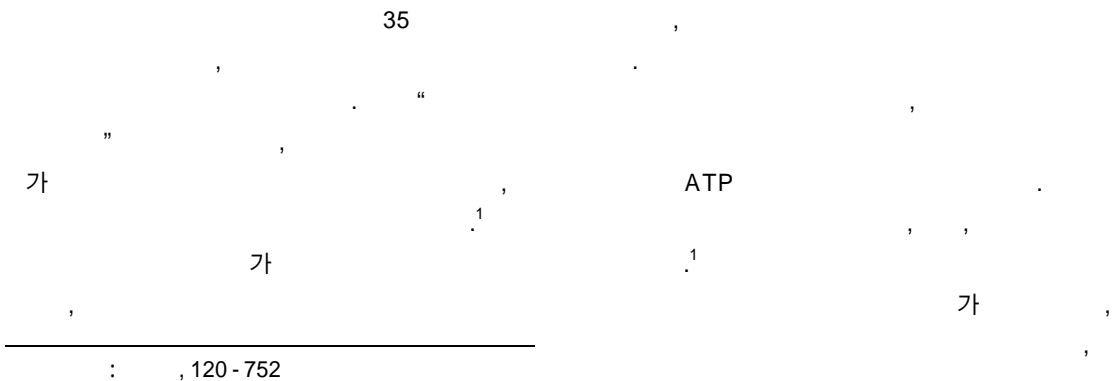
Kyoung Min Choi, M.D.,<sup>1</sup> Hae Sik Kweon, M.D.,<sup>1</sup> Dong Woo Lee, M.D.,<sup>1</sup>  
Ran Namgung, M.D.,<sup>1</sup> Min Soo Park, M.D.,<sup>1</sup> Chul Lee, M.D.,<sup>1</sup>  
Young Mok Lee, M.D.,<sup>2</sup> Jin Sung Lee, M.D.<sup>2</sup>

Departments of Pediatrics,<sup>1</sup> Clinical Genetics,<sup>2</sup> College of Medicine, Yonsei University, Seoul, Korea

Mitochondrial diseases are classified into the three major categories, defects of fatty acid oxidation, defects of pyruvate metabolism, and defects of the respiratory chain, and all of these cause severe neurologic dysfunction in the newborn period. Defects of the mitochondrial respiratory chain present as recurrent apnea, seizures, congenital lactic acidosis, hypotonia, hepatic dysfunction and hypertrophic cardiomyopathy in the neonatal period. Laboratory findings of hyperlactataemia (>2.5mM), elevated lactate/pyruvate (L/P) ratio (>20) and ketone body ratio (>2) suggest the diagnosis of mitochondrial respiratory chain defects.

We report a case of mitochondrial respiratory chain defect diagnosed in the neonatal period presenting with multiorgan failure consisting of severe metabolic acidosis, comatous mental state, respiratory distress, hepatic dysfunction, renal failure with lactic acidosis (24mM), increased L/P ratios (55.6) and ketonuria (increased ratio of 3-hydroxybutyrate/acetacetate).

**Key words :** Mitochondrial respiratory chain defect, Lactic acidosis, Newborn.



: , 120 - 752  
(02) 361 - 5510 · (02) 393 - 9118  
E - mail : ranng@yumc.yonsei.ac.kr

1  
 ratios 가( >20, 가(>2.5mM), molar  
 1  
 100 36% , 44% 1 2  
 , 20% 2  
 2



Fig. 1. General appearance of patient.

(>2.5mM), 가 /  
 (>20),  
 1

(Fig. 1),

**증 례**

: , 17 , .  
 :  
 가 : 39 5  
 3,240gm(25~  
 50 ), 58cm(97 ),  
 37cm(90~97 ) , 가 ,  
 .가 가  
 :  
 : 7 3  
 가  
 BCG 가  
 :  
 66/39mmHg,  
 36.7 , 160 / ,  
 , 3,230gm .

가 .  
 (Fig. 1),  
 가 .  
 : 2  
 가 . Babinski  
 : pH 6.6, PaCO<sub>2</sub> 16.3mmHg,  
 PaO<sub>2</sub> 186.8mmHg, - 33mM, 97%  
 (anion gap) 48.4  
 19,600/mm<sup>3</sup>(  
 37%, 58%, 1%), 11.7g/dL,  
 604,000/mm<sup>3</sup> , Na<sup>+</sup> 146  
 mEq/L, K<sup>+</sup> 6.4mEq/L, Cl<sup>-</sup> 90mEq/L, HCO<sub>3</sub><sup>-</sup> 5mEq/L,  
 174mg/dL . Na<sup>+</sup> 11  
 170mEq/L 가 . BUN/  
 Cr 50/2.8mg/dL, SGOT/SGPT 508/257IU/L,  
 584 μg/dL 가  
 . PT 30%, PTT 70.1

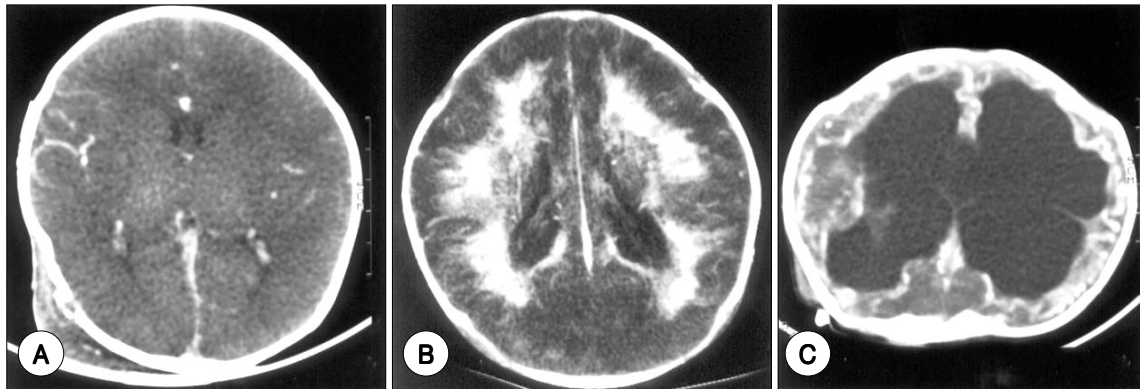


Fig. 2. A : Hospital day 1, B : Hospital day 25, C : Hospital day 90, Brain CT shows progression of parenchymal destruction with hydrocephalus.

, FDP>1 : 64 , D - dimer  
 , fibrinogen 105mg/dL  
 EEG  
 : 1  
 10  
 . 90  
 (Fig. 2).  
 20 pH 7.58, - 6.8mM  
 22 40 µg/dL  
 foy  
 phenobarbital, phenytoin va-  
 lproic acid  
 가 48.4  
 2.2mmol/L), 0.424mmol/L( 0.03~  
 0.08mmol/L) , / 가 55.6  
 20 가 .

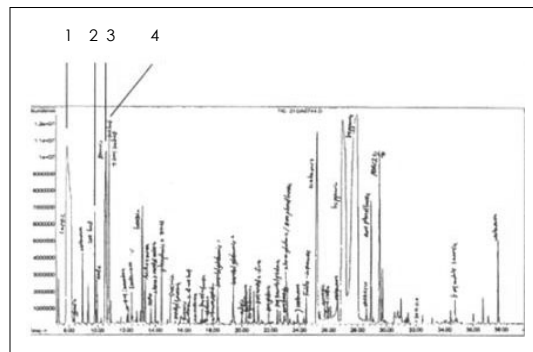


Fig. 3. Urine organic acid assay shows massive elevations of lactate, pyruvate and 2-OH butyrate with severe ketonuria(3-hydroxybutyrate). 1, lactate : 2, 2-OH butyrate : 3, pyruvate : 4, 3-hydroxybutyrate.

(3 - hydroxybutyrate/acetoacetate ratio 가)  
 , 2 - OH butyrate 가 (Fig. 3),  
 2,237nmol/mL(  
 : 131.0 - 710.0nmol/mL) 가  
 20mg/  
 day, 20mg/day, 400mg/day,  
 coenzyme Q 30mg/day, L - carnitine 1.6g/day

. BUN/Cr  
 , SGOT/SGPT PT/PTT  
 . BUN/Cr 16 18.8/1.0 ,

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SGOT/SGPT 8 59/43 IU/L  
 flat EEG 가 / 가<sup>6</sup>  
 (oculocephalic movement)가  
 , calory (caroric - indu-  
 ced eye movement)가  
 가  
 가  
 가  
 가 4<sup>7,8</sup>

**고 찰**

1987 1996  
 4.7/100,000<sup>3</sup>  
 roof  
 가 (nu-  
 molar ratio가 clear DNA mitochondrial DNA)  
 (>2.5mM), 가 / (>20), 가<sup>5</sup>  
 가 molar ratio(>2) replicative segregation  
 가<sup>5</sup>  
 가 , Re-  
 , ATP plicative segregation 가  
 가<sup>5</sup>  
 가<sup>4</sup>  
 가 , DNA(mtDNA) 가  
 가 mtDNA 가  
 가  
 mtDNA 가  
 가  
 (oxidation - reduction potential)가  
 가 / 가 20  
 가  
 가 /  
 , cytochrome C oxidase<sup>9</sup>  
 , 1

rate) 10~20 가 (mutation fixation DNA (2~4g/day) 가 , mtDNA , mtDNA , mtDNA , 80% , sodium valproate tetracycline chloramphenicol 가 가 Vit K<sub>3</sub>(40~160mg/day), coenzyme Q<sub>10</sub>(80~300mg/day)

가 , complex (100mg/day) 가

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