4.7 Mass Screening of Congenital Metabolic Disorder (Phenylketonuria) (1) - GCMS

•Explanation

With mass screening of congenital metabolic disorder, GC/MS identifies and quantifies ground substances and erroneous metabolic elements that have accumulated due to a disorder in a certain metabolic route in order to locate the metabolic region and enable diagnosis of the disease.
Phenylketonuria is an ailment where the symptoms - including central nervous disorders such as intellectual impairment, convulsions and brain wave errors, and melamine achromia - are determined from the latter half of infancy.
Fig. 4.7.1 shows the metabolic route of phenylalanine. As Fig. 4.7.2 shows, phenylketonuria can be diagnosed if phenylpyruvic acid, phenyllactic acid and phenylalanine can be found in the urine sample.

•Analytical Conditions

Instrument	: GCMS-QP5000
– GC –	
Column	: DB-5(30m × 0.25mmi.d. df=0.25µm)
Column Temp.	: 60˚C – 17˚C/min – 325˚C(3min)
Injector Temp.	: 260˚C
Carrier gas	: He 50kPa
Injection Method	: Split (20:1)
– MS –	
Interface Temp.	: 280˚C
Ionization Method	: EI
Scan Range	: m/z 50-650(EI)
Scan Interval	: 0.4sec

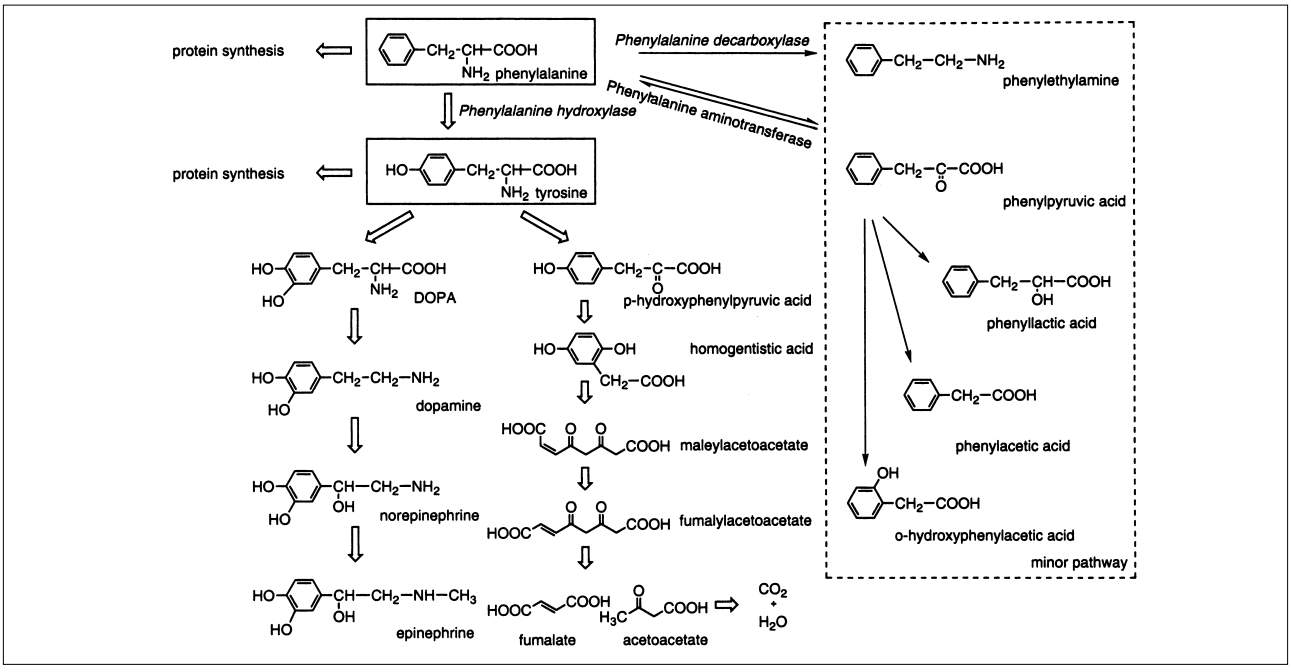

Fig. 4.7.1 Phenylalanine metabolic route

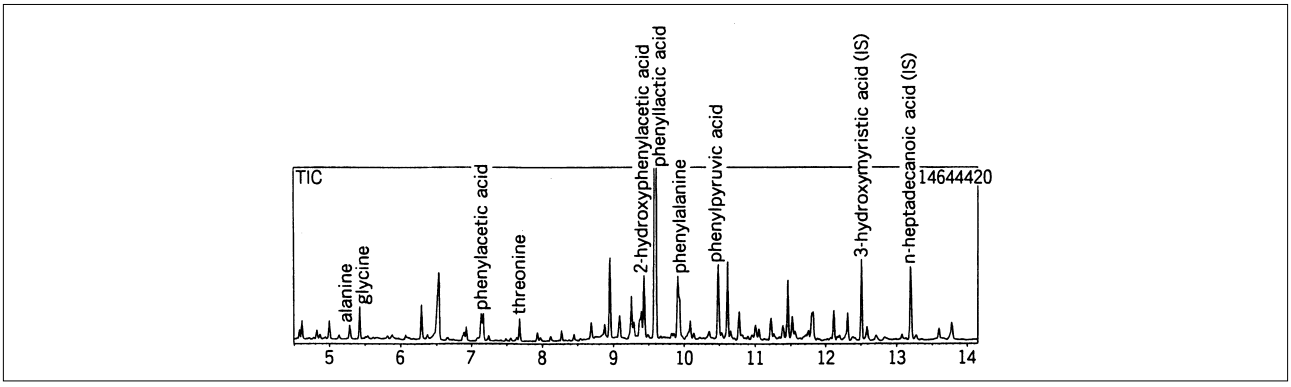

Fig. 4.7.2 Total ion chromatogram of urine from a phenylketonuria patient

4.7 Mass Screening of Congenital Metabolic Disorder (Propionic Acidemia and Methylmalonic Acidemia) (2) - GCMS

•Explanation

With mass screening of congenital metabolic disorder, GC/MS identifies and quantifies ground substances and erroneous metabolic elements that have accumulated due to a disorder in a certain metabolic route in order to locate the metabolic region and enable diagnosis of the disease.

Propionic acidemia and methylmalonic acidemia is recognized by symptoms such as vomiting, lethargy, myotony reduction and growth retardation.

Fig. 4.7.3 shows the metabolic route of isoleucine. As Fig. 4.7.4 shows, propionic acidemia can be diagnosed if 3-hydroxyisovaleric acid, tiglylglicine and methylcitiric acid can be found in the urine sample. As Fig. 4.7.5 shows, methylmalonic acidemia can be diagnosed if methylmalonic acid and 3-hydroxypropionic as a micro metabolic substance can be found in the urine sample.

•Analytical Conditions

Instrument	: GCMS-QP5000
– GC –	
Column	: DB-5(30m × 0.25mmi.d. df=0.25μm)
Column Temp.	: 60˚C – 17˚C/min – 325˚C(3min)
Injector Temp.	: 260˚C
Carrier gas	: He 50kPa
Injection Method	: Split (20:1)
– MS –	
Interface Temp.	: 280˚C
Ionization Method	: EI
Scan Range	: m/z 50-650 (EI)
Scan Interval	: 0.4sec

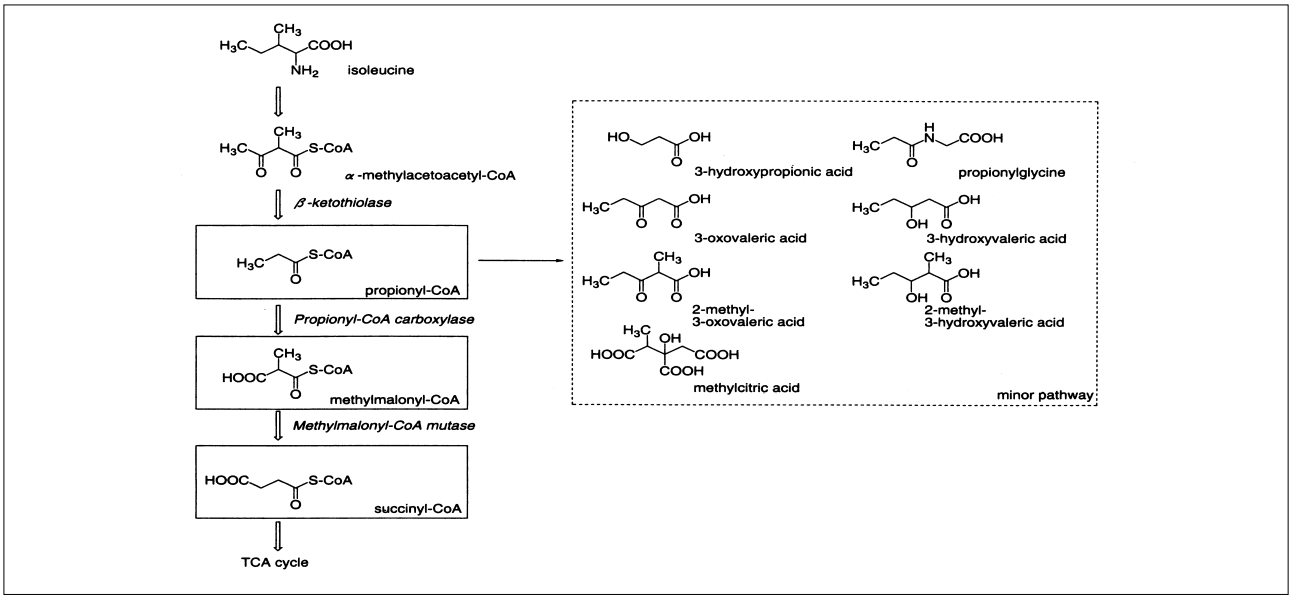

Fig. 4.7.3 Isoleucine metabolic route

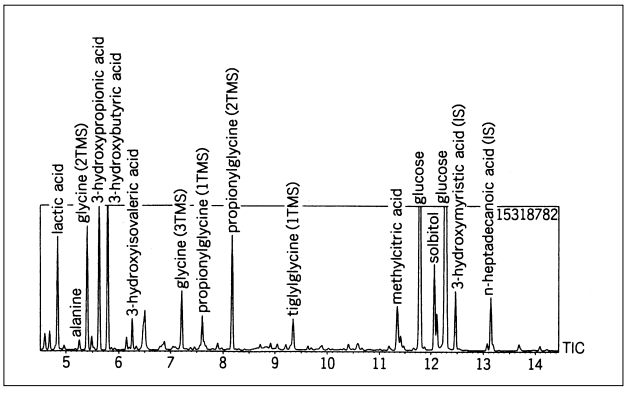

Fig. 4.7.4 Total ion chromatogram of urine from a propionic acidemia patient

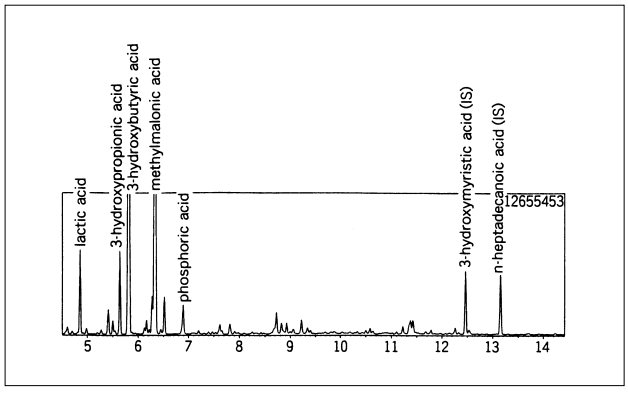

Fig. 4.7.5 Total ion chromatogram of urine from a methylmalonic acidemia patient

4.7 Mass Screening of Congenital Metabolic Disorder (Isovaleric Acidemia) (3) - GCMS

•Explanation

With mass screening of congenital metabolic disorder, GC/MS identifies and quantifies ground substances and erroneous metabolic elements that have accumulated due to a disorder in a certain metabolic route in order to locate the metabolic region and enable diagnosis of the disease.

Isovaleric acidemia is an ailment with symptoms including the generation of an odor that smells like "sweaty feet", polypnea, vomiting and lowering of consciousness. This ailment can lead to death if untreated.

Fig. 4.7.6 shows the metabolic route for leucine. As shown in Fig. 4.7.7, isovaleric acidemia can be diagnosed if isovalerylglycine and 3-hydroxyisovaleric acid can be found in the urine sample.

•Analytical Conditions

Instrument	: GCMS-QP5000
– GC –	
Column	: DB – 5(30m × 0.25mmi.d. df=0.25μm)
Column Temp.	: 60˚C – 17˚C/min – 325˚C(3min)
Injector Temp.	: 260˚C
Carrier gas	: He 50kPa
Injection Method	: Split (20:1)
– MS –	
Interface Temp.	: 280˚C
Ionization Method	: EI
Scan Range	: m/z 50-650 (EI)
Scan Interval	: 0.4sec

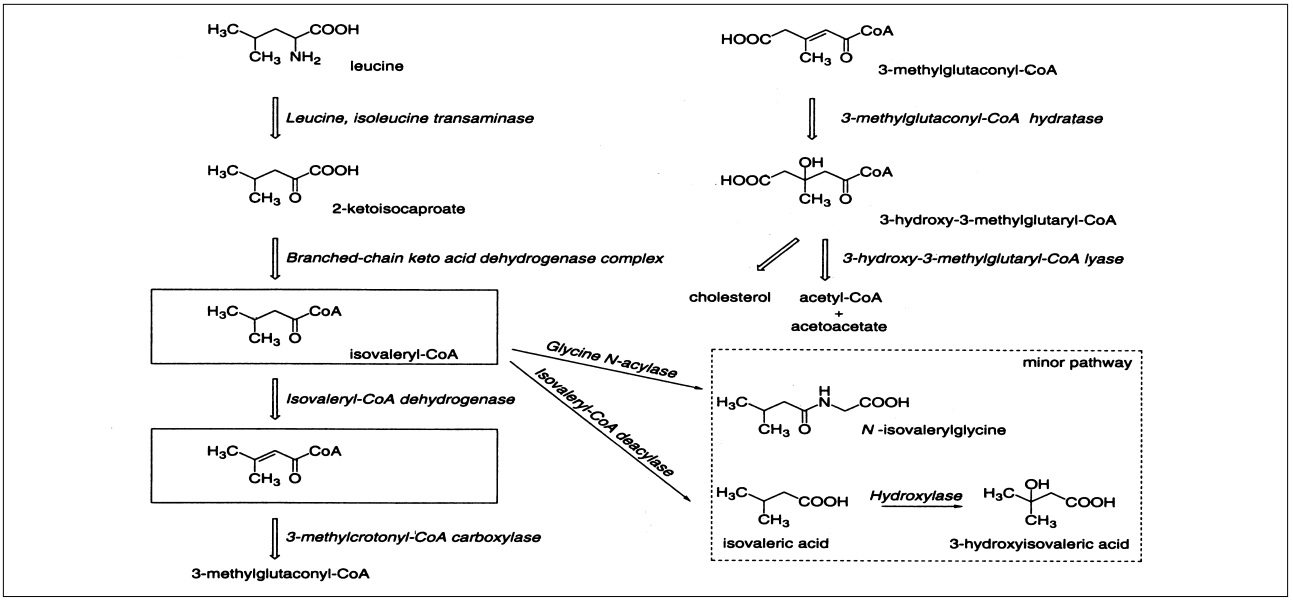

Fig. 4.7.6 Leucine metabolic route

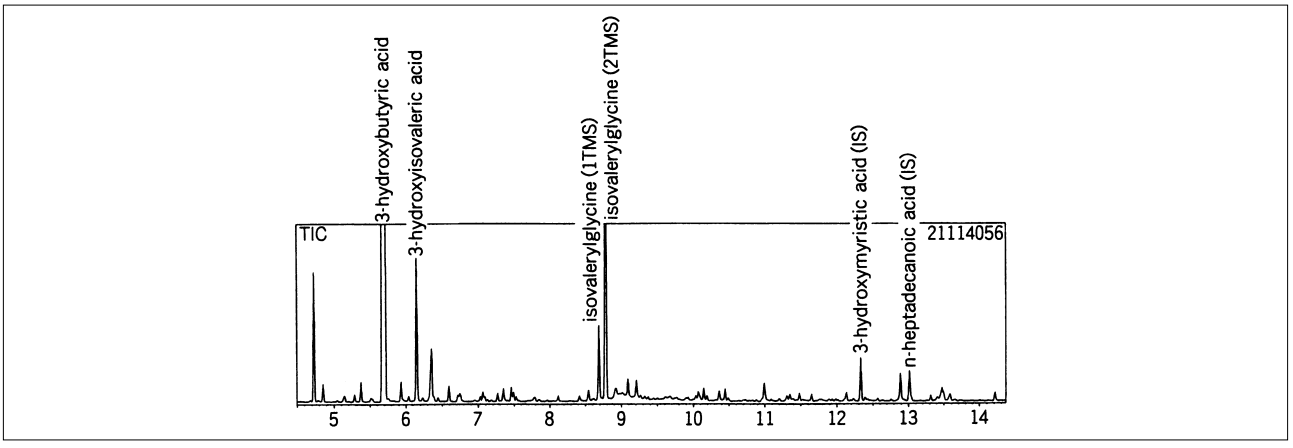

Fig. 4.7.7 Total ion chromatogram of urine from isovaleric acidemia patient