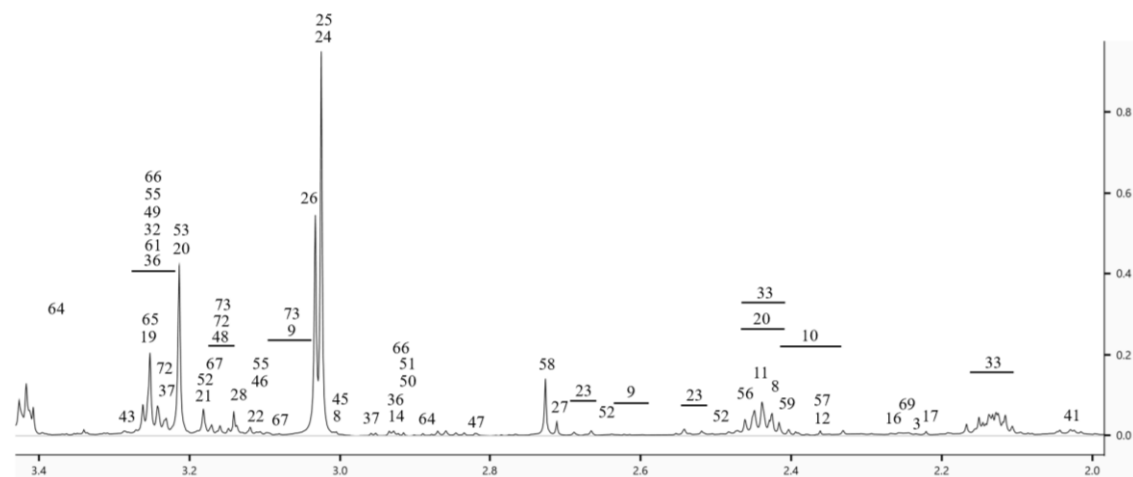
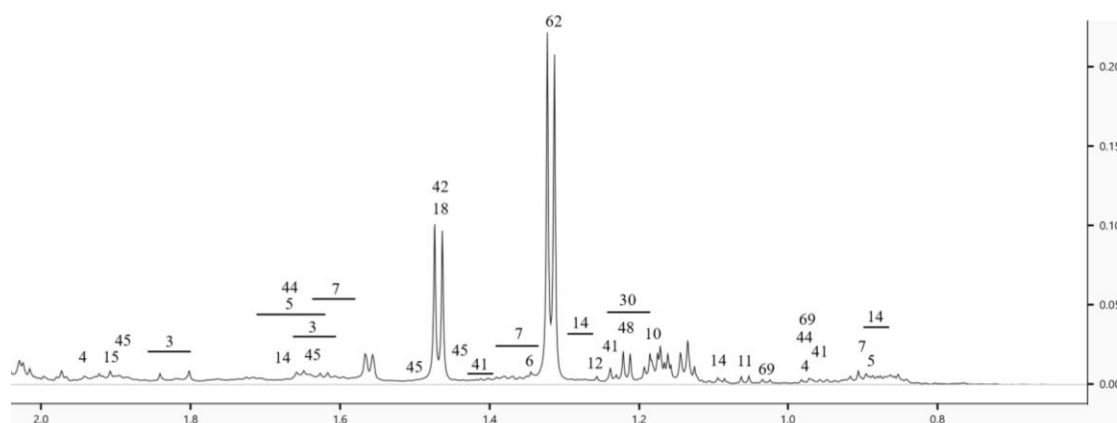
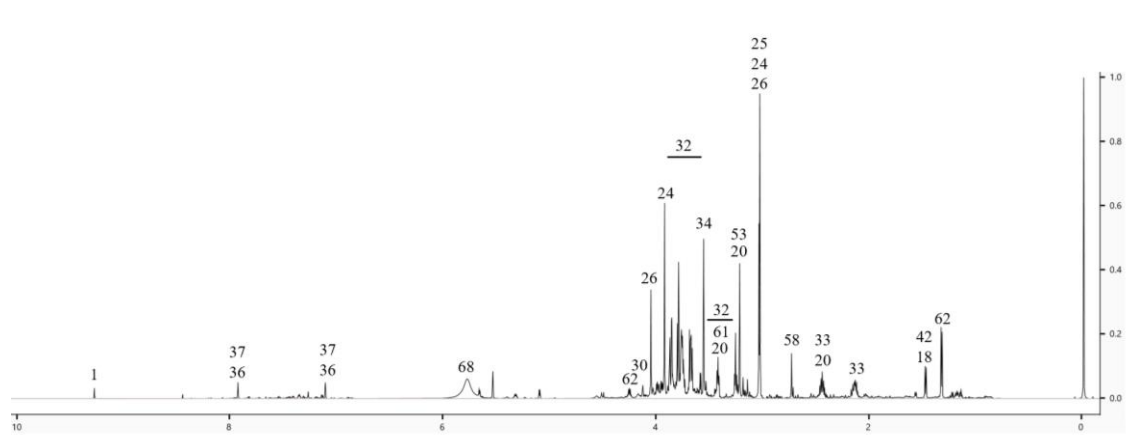
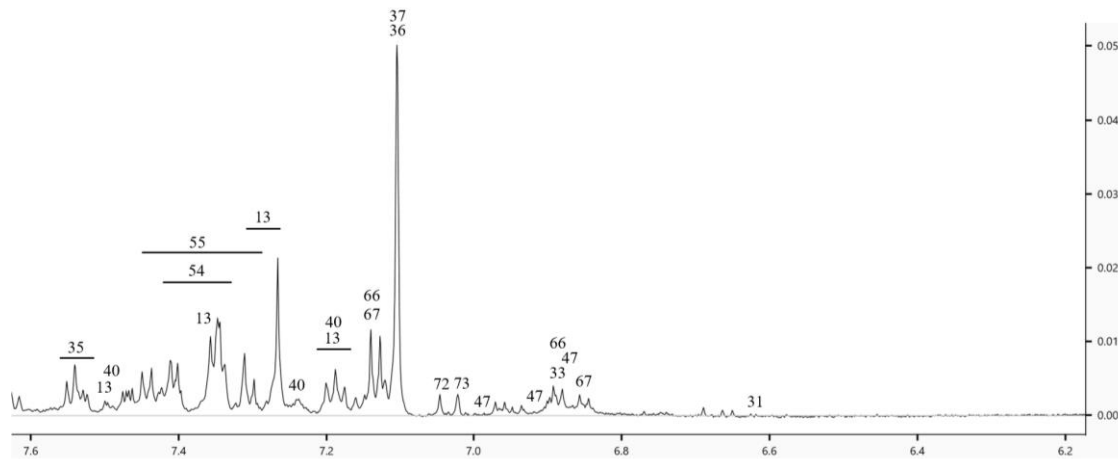
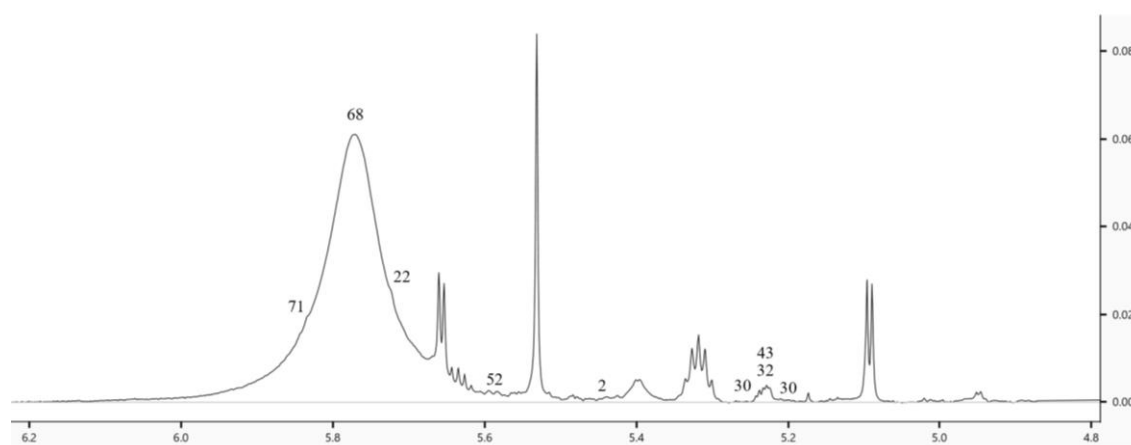
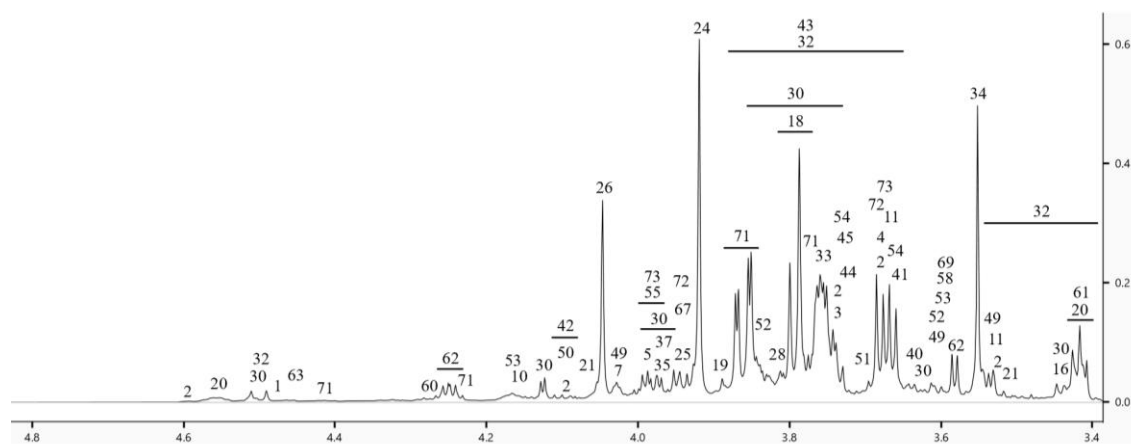
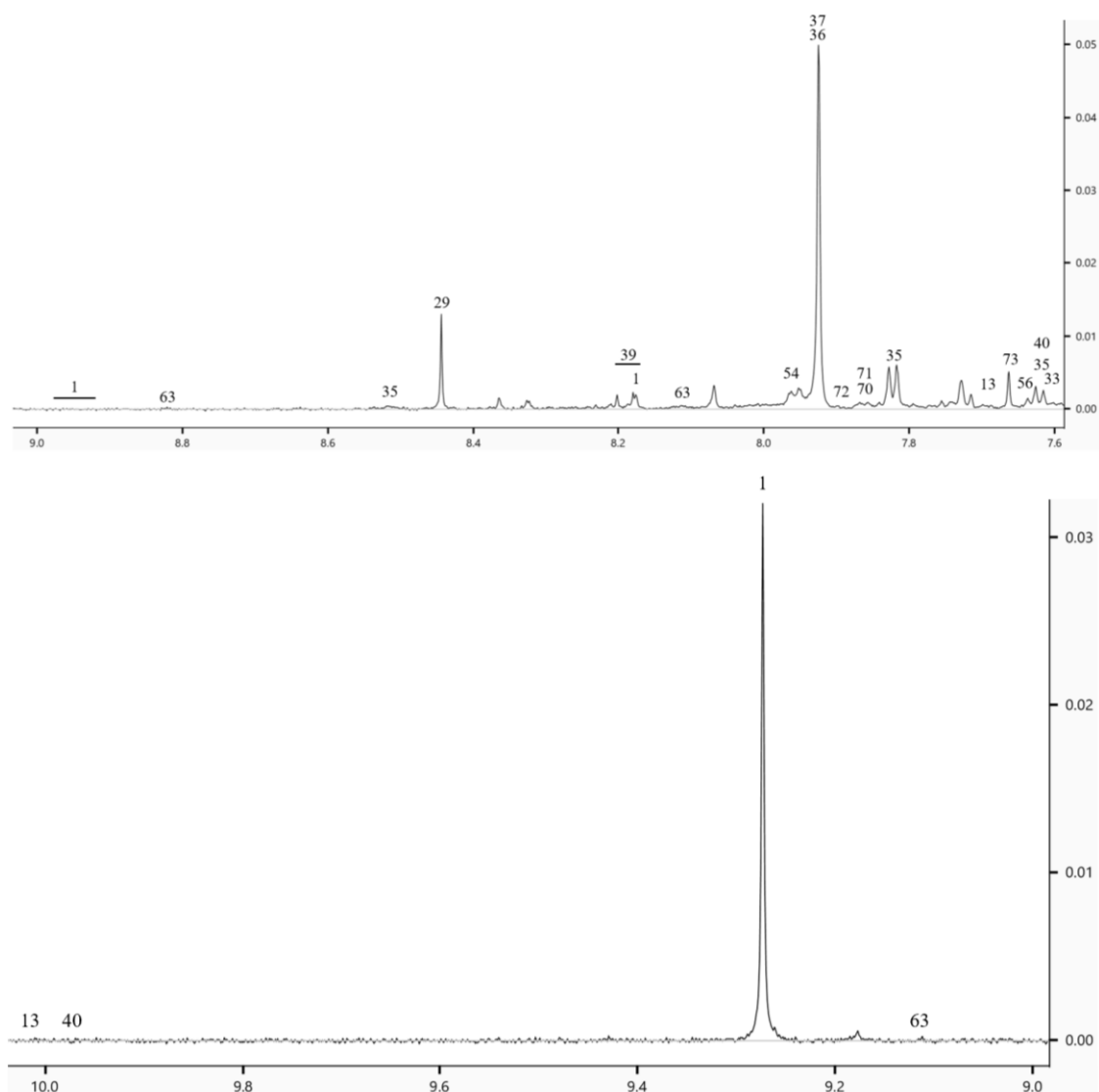








Additional File 2. Labeled NMR spectrum of patient 45 using Chenomx. Includes the 73 identified metabolites listed in Table 2. Concentration values obtained relative to the internal standard TSP. X-axis is chemical shift in parts per million (ppm). Peaks are marked by metabolite number: 1) 1-methylnicotinamide, 2) 1,6-Anhydro-beta-D-glucose, 3) 2-aminoadipate, 4) 2-aminobutyrate, 5) 2-hydroxybutyrate, 6) 2-hydroxyisobutyrate, 7) 2-hydroxyvalerate, 8) 2-oxoglutarate, 9) 3-aminoisobutyrate, 10) 3-hydroxybutyrate, 11) 3-hydroxyisobutyrate, 12) 3-hydroxyisovalerate, 13) 3-indoxylsulfate, 14) 3-methyl-2-oxovalerate, 15) acetate, 16) acetoacetate, 17) acetone, 18) alanine, 19) betaine, 20) carnitine, 21) choline, 22) cis-aconitate, 23) citrate, 24) creatine, 25) creatine phosphate, 26) creatinine, 27) dimethylamine, 28) ethanolamine, 29) formate, 30) fucose, 31) fumarate, 32) glucose, 33) glutamine, 34) glycine, 35) hippurate, 36) histamine, 37) histidine, 38) histamine, 39) hypoxanthine, 40) indole-3-acetate, 41) isoleucine, 42) lactate, 43) lactose, 44) leucine, 45) lysine, 46) malonate, 47) methylguanidine, 48) methylmalonate, 49) myo-inositol, 50) n-methylhydantoin, 51) n,n-dimethylglycine, 52) o-acetylcarnitine, 53) o-phosphocholine, 54) phenylacetylglycine, 55) phenylalanine, 56) pyridoxine, 57) pyruvate, 58) sarcosine, 59) succinate, 60) tartrate, 61) taurine, 62) threonine, 63) trigonelline, 64) trimethylamine, 65) trimethylamine n-oxide, 66) tyramine, 67) tyrosine, 68) urea, 69) valine, 70) xanthine, 71) xanthosine, 72) pi-methylhistidine, 73) tau-methylhistidine.