

CMA Case 2

A 4-year-old girl with metabolic acidosis and increased bone density

A 4-year-old girl had an uncomplicated perinatal course, but during infancy she was found to have failure to thrive, metabolic acidosis, and global developmental delay. She was diagnosed as having renal tubular acidosis. A chest X-ray showed generalized increased bone density. Her medical history is also significant for asthma.

Family history: Parents are first cousins and unaffected. She had three healthy brothers and one similarly affected sister.

Regions of homozygosity **using hg19 assembly**

chr1:28,213,441-55,046,130
chr2:233,855,461-237,405,689
chr3:122,533,517-131,755,403
chr4:40,722,751-44,414,165
chr4:153,249,146-157,441,782
chr5:123,878-10,020,635
chr8:16,696,291-26,667,066
chr8:62,924,308-96,465,727
chr9:108,011,356-113,737,232
chr12:28,701,096-41,656,532
chr12:96,057,387-104,974,514
chr14:63,464,096-70,381,073
chr16:5,073,773-8,731,200
chr16:61,155,221-73,033,473
chr18:58,397,130-65,752,002
chr20:124,302-3,972,793
chr20:40,809,746-49,922,735