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Multiple Nephrocalcinosis in an Infant with 3p Deletion Syndrome

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Case Study:

The 3p deletion syndrome is a rare disorder that results from a chromosomal change and deletion of the end of the small (p) arm of chromosome 3. The size of the deletion varies among affected individuals, ranging from approximately 150,000 DNA building blocks (150 kilobases or 150 kb) to 11 million DNA building blocks (11 megabases or 11 Mb). Most cases of 3p deletion syndrome are not inherited but de novo. The deletion occurs in one chromosome, most often as a random event during the formation of reproductive cells (eggs or sperm) or in early fetal development. In these cases, affected people have no history of the disorder in their family.

3p deletion syndrome is characterized by growth retardation, developmental delay, mental retardation, dysmorphism, microcephaly, and ptosis. Additionally, individuals with 3p deletion syndrome may have seizures, weak muscle tone (hypotonia), intestinal abnormalities, or congenital heart defects. The phenotype of individuals with deletions varies from normal to severe. The variable penetrance of 3p deletions creates challenges in genetic counseling, as the phenotype of the offspring cannot be predicted based on chromosomal and/or genome-wide array analytical findings.

Little is known about a meaningful association between the 3p deletion and nephrocalcinosis. Here, we describe a 50-day-old infant with multiple nephrocalcinosis in both kidneys and chromosome 46, XY, del(3)(p25-pter).