CASE REPORT

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Sotos Syndrome and Nephrocalcinosis a Rare But Possible Association Due to Impact on Contiguous Genes

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What is already known on this topic?

Up to 15% of patients with Sotos syndrome present with renal disorders, the most common of which is vesicoureteral reflux. However, nephrocalcinosis is a less common clinical disorder reported in cases of Sotos syndrome due only to deletion that includes *SLC34A* and other genes, associated with phosphate wasting and hypercalciuria.

What this study adds?

In this patient group, delayed growth and impaired kidney function are possible and long-term follow-up is recommended. The exceptional circumstance presented in this brief report are the presentation and long-term follow-up of a patient with genetically confirmed Sotos Syndrome in association with a less common clinical disorder, nephrocalcinosis. This association can be explained by the alteration of contiguous genes included in the deletion found in 5q35 and the unusual fibroblast growth factor 23 values.

Abstract

One-month old, breastfeeding infant, born at term, with normal anthropometric measurements at birth was referred to Pediatric Nephrology due to a nephrocalcinosis. The patient presented with dysmorphic features and heart disease. A metabolic study was conducted on blood and urine yielding results within normal parameters, except for the renal concentration test and acidification test. At six months of age, the patient presented with overgrowth, which along with other clinical signs aroused the suspicion of Sotos syndrome. Molecular genetic testing identified a heterozygous deletion in 5q35 between bands q35.2 and q35.3, affecting the genes *NSD1, SLC34A1* and *FGFR4*, which was compatible with Sotos syndrome and with nephrocalcinosis as a rare association.

Keywords: Sotos syndrome, nephrocalcinosis, *NSD1, SCL34A1, FGFR4*, case report

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Introduction

Sotos syndrome is a dominant, autosomal, hereditary disease with a prevalence of approximately 1 in 14,000 newborns (1,2). It is characterized by overgrowth, a distinctive facial phenotype and learning disabilities (3). Sotos syndrome is a multisystemic disorder and up to 15% of patients present with renal disorders, the most common of which is vesicoureteral reflux. Most cases of Sotos syndrome are found to harbor point mutations in *NSD1* gene and these are *de novo* in over 95% of cases. Clinical findings in European patients show 10-15% microdeletion of 5q35 affecting this gene, as found in the presented case discussed herein (2). The exceptional circumstance in this patient is the association of genetically confirmed Sotos syndrome in association with a less common clinical disorder, nephrocalcinosis. This association may be explained by the alteration of contiguous genes included in the deletion found.

Case Report

A 37-day old, male, breast-feeding infant was referred to pediatric nephrology due to a finding of bilateral, medullary nephrocalcinosis on an ultrasound examination conducted at one month of life. The patient was born from a well-mamaged pregnancy without incident, with normal prenatal echographic findings, from non-consanguineous parents and with no relevant family history. Labor at 42 weeks of pregnancy, concluded by C-section due to failed induction, with adequate weight at birth of 3900 g [+0.93 standard deviation (SD) score (SDS)]. The infant was admitted to the neonatal unit at 3 hours of life due to hypoglycemia, developed histologically-confirmed eosinophilic colitis symptoms at eight days of life and exhibited good progress after 10 days of parenteral feeding. During his hospital stay, the patient received aminoglycoside treatment.

The infant was monitored in pediatric cardiology due to *ostium secundum* atrial septal defect without hemodynamic compromise, patent ductus arteriosum (PDA) and mild pulmonary valvular stenosis, and in pediatric neurology due to mild hypotonia and subtle dysmorphic features (broad forehead, lack of frontotemporal hair, long and narrow face, single transverse palmar crease, scaphocephaly and anteverted nares). There was also a front left paraventericular ependymal cyst and FLAIR hyperintensity in the corona radiata on brain magnetic resonance imaging. Stimulation was begun in early care due to mild neurodevelopmental retardation. Neonatal metabolic screening was compatible with normal characteristics and otoacoustic emissions yielded negative results. He received a daily supplement of vitamin D_3 (400 IU) and omeprazole. Normal ophthalmological examination was reported.

Given the presence of bilateral nephrocalcinosis and the patient's history, he was investigated for an initial differential diagnosis of a probable syndrome, a renal condition and/or a multi-factor etiology due to associated neonatal comorbidity. A metabolic study was conducted on blood and urine with normal results, including thyroid hormones, lactate, pyruvate and organic acids, although a gradual decrease of blood phosphate levels was observed, reaching values even lower than the reference values for the patient's age, with phosphate reabsorption and normal fibroblast growth factor 23 (FGF23) values which were inappropriate for phosphatemia. Of note, he also exhibited an increase in 1,25 $(OH)_2$ vitamin D_3 and parathyroid hormone (PTH) was in the low-normal range, appropriate for his serum calcium levels which were at the upper end of the normal range (Table 1). He had normal proteinuria results, including tubular proteinuria. Initially, renal function showed an increase in cystatin C and N-acetylglucosaminidase (NAG) values, as well as abnormal renal concentration test results after stimulation with desmopresin, reaching maximum urinary osmolality of 307 mOsm/Kg (normal value >562 mOsm/Kg), as well as altered renal acidification capacity, reaching a maximum pCO_2 of 54 mmHg after stimulation with bicarbonate and acetazolamide (normal value >70 mmHg).

At four months of age, he presented again with *Escherichia coli* urinary tract infection with fever, with normal cystourethrogram, while the renal gammagraphy showed right kidney hypodysplasia with no associated cortical lesions. At six months, overgrowth was detected with weight +2.50 SDS, height +2.63 SDS and head circumference +2.38 SDS. However, the anthropometric values gradually return to normal. Furthermore, bone age corresponded to patient's age and no associated skeletal disorders were observed.

Sotos syndrome was suspected due to clinical observations of dysmorphic syndrome, overgrowth and heart disease. Given the association with nephrocalcinosis, an expanded genetic study was requested to search for microdeletions that affect genes other than *NSD1* due to contiguity. The fluorescence in situ hybridization analysis revelaed an absence of hybridization signal in one of the loci for Sotos syndrome on chromosome 5, with a normal pattern in the parents (Figure 1). In addition, oligo-comparative genomic hybridization (CGH) array confirmed a *de novo* deletion of approximately 2 Mb in 5q35.2 to q35.3 (from 175,580,042 to 177,386,153 bp) in the patient, as oligo-CGH array using maternal versus paternal DNA yielded a

Table 1. Analytical data of the patient over time

Age	1 m	2 y	4 y	6 y	7 y
Plasma (reference values in parentheses)					
Cr (mg/dL) (0.32-0.59)	0.34	0.34	0.46	0.59	0.55
Cystatin C (mg/L) (0.62-1.11)	2	0.62	1.02	0.96	-
Urate (mg/dL) (2.2-4.5)	2.4	2.7	-	3.1	2.6
Ion Ca (mg/dL) (4.6-5.3)	5.8	5.5	5	5.3	-
Phosphate (mg/dL) (4.1-5.9)	5.3	3.9	3.9	3.2	4
Magnesium (mg/dL) (1.6-2.6)	2.1	2.0	1.8	1.8	1.8
AP (U/L) (142-335)	500	335	302	379	319
PTH (pg/mL) (11-60)	14	24	14	15	14
25 (OH) Vit D3 (ng/mL) (17-49)	-	42	39	33	39
1,25 $(OH)_2$ Vit D3 (pg/mL) (45-102)	-	249	-	99	-
Intact FGF23 (pg/mL) (36 ± 18)	-	51.3	-	-	-
pH_p (7.35-7.45)	-	7.37	7.39	7.37	7.38
Bicarbonate (mEq/L) (20-26)	-	25	23	22	23
Urine (reference values in parentheses)					
pH_u	7.5	7	7	7	7.5
Ca/Cr (mg/mg) (< 0.20-0.28)	0.52	0.35	0.28	0.26	0.25
Citrate/Cr (mg/g) (> 250-420)	706	687	307	172	314
Ca/citrate (mg/mg) (< 0.33)	0.74	0.51	0.92	1.49	0.58
Oxalate/Cr (mg/g) (< 110)	130	60	50	-	41
Prot/Cr (mg/mg) (< 0.2)	0.68	0.18	0.18	0.11	0.11
NAG/Cr (U/g) (< 6-11)	150	15	-	3	3
TRP (mg/dL GFR) (91.05 ± 4.71)	91	90	79	80	88
TP/GFR (mg/dL) (4.6 ± 0.6)	4.8	3.62	3.1	2.3	3.5
V/GFR (mL/dL GFR) (0.59 ± 0.22)	1.44	1.10	1.20	0.91	0.98

m: month, y: years, Cr: creatinine, Ca: calcium, AP: alkaline phosphatase, PTH: parathyroid hormone, 25(OH) vit D_3: 25-hydroxyvitamin D_3, 1,25$(OH)_2$ vit D_3: 1,25-dihydroxyvitamin D_3, pHp: plasma pH, pHu: urine pH, Prot/Cr: protein/creatinine ratio, TRP: tubular reabsorption of phosphate, TP/GFR: tubular reabsorption of phosphate per dL of glomerular filtrate, V/GFR: urinary volume per dL of glomerular filtrate

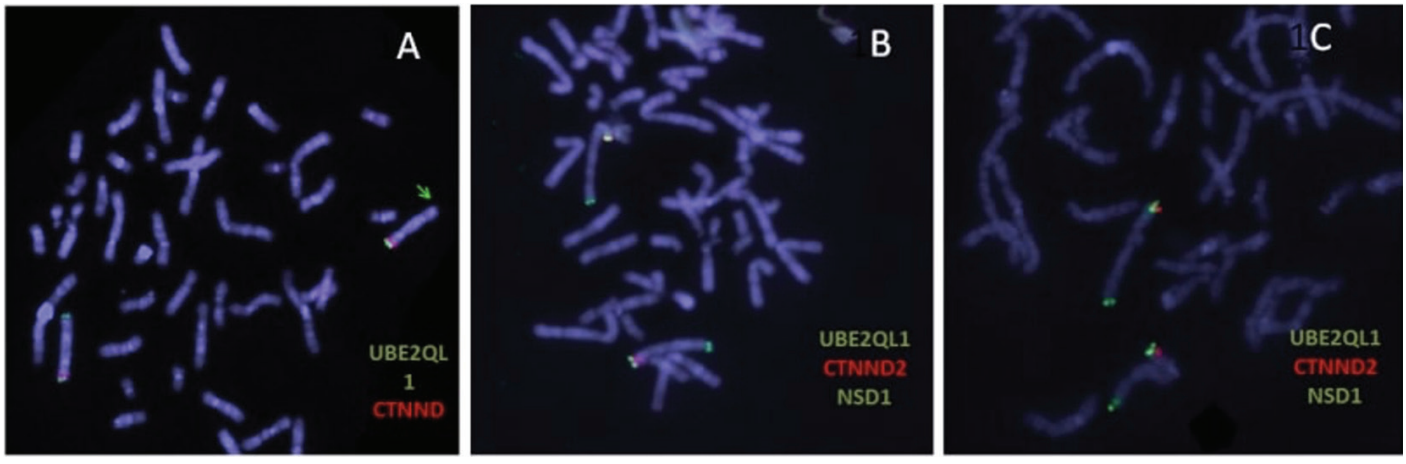

Figure 1. (A) FISH of the patient with probe combined for the loci of the Cri-du-chat (*UBE2QL1*, 5p15.31; *CTNND2*, 5p15.2) and Sotos (*NSD1*, 5q35) syndromes. The signals in 5p15 are present in both chromosome 5 pairs. The arrow shows lack of hybridization of the *NSD1* clone (green) in one of the chromosome 5 pairs. B, C) FISH of the father and mother, respectively, with the same probe. Both chromosome 5 pairs show signals both in 5p15 and 5q35

FISH: fluorescence in situ hybridization

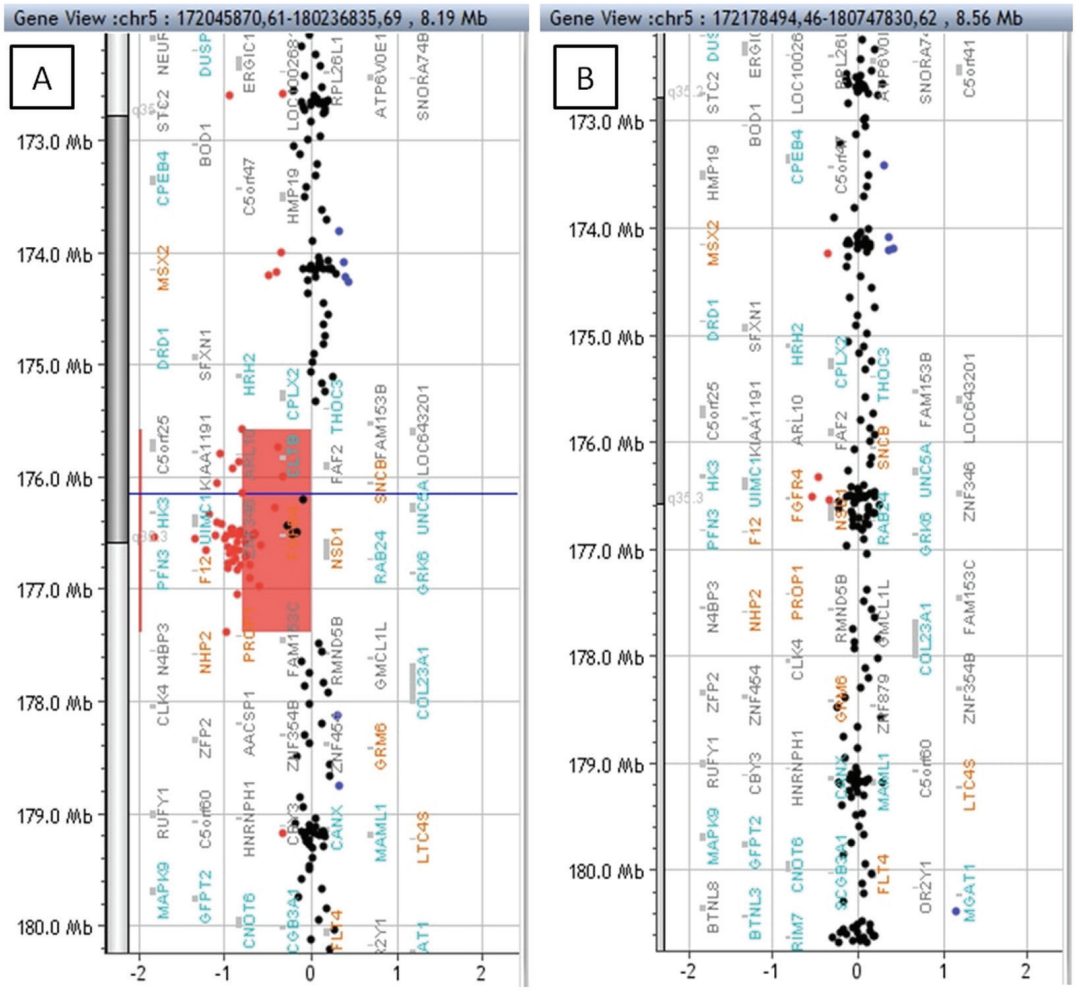

Figure 2. (A) Oligo-array CGH of the patient, showing an enlargement of the deleted region (red bar) in chromosoma 5. B) Oligo-array CGH of the same region in chromosome 5 after comparing the DNA of the parents, illustrating a normal result

CGH: comparative genomic hybridization

normal hybridization profile, including the genes *SLC34A1* and *FGFR4* in this region, among others (Figure 2).

At four years of age, oral phosphate was prescribed, with poor tolerance, so he finally began thiazides due to improvement in serum calcium levels. In the last assessment at seven years of age, weight and height were normal at -0.07 SDS and +0.44 SDS, respectively, with normal growth rate (-0.7 SD) and insulin-like growth factor-1 (IGF-1) and IGF binding protein 3 values within the normal range for the patient's age. Bilateral medullary nephrocalcinosis persisted, with renal asymmetry (right kidney, 50-75th percentile and left kidney >95th percentile), and the abnormal test results shown in Table 1, along with mild hypercalciuria. However, there was an improvement in phosphate levels and normalization of renal function tests. Cardiological evaluation showed continuation of mild PDA without other findings. In terms of neurological development, the patient continues to receive cognitive and communication stimulation and his psychomotor retardation was improving. Cow's milk protein challenge was undertaken and tolerance was good.

Discussion

The *NSD1* gene is the only gene currently known to cause Sotos syndrome. Among European patients with typical findings of this syndrome, up to 15% present 5q35 microdeletion that affects NSD1, and associated disorders may appear when these deletions affect other genes (2,4). There is no genotype-phenotype correlation in this syndrome, but in cases due to 5q35 microdeletion, overgrowth is less obvious and usually has more neurological impact, as is the case in our patient, while nephrocalcinosis is a phenotypic characteristic reported only in cases of Sotos syndrome due to microdeletion (4).

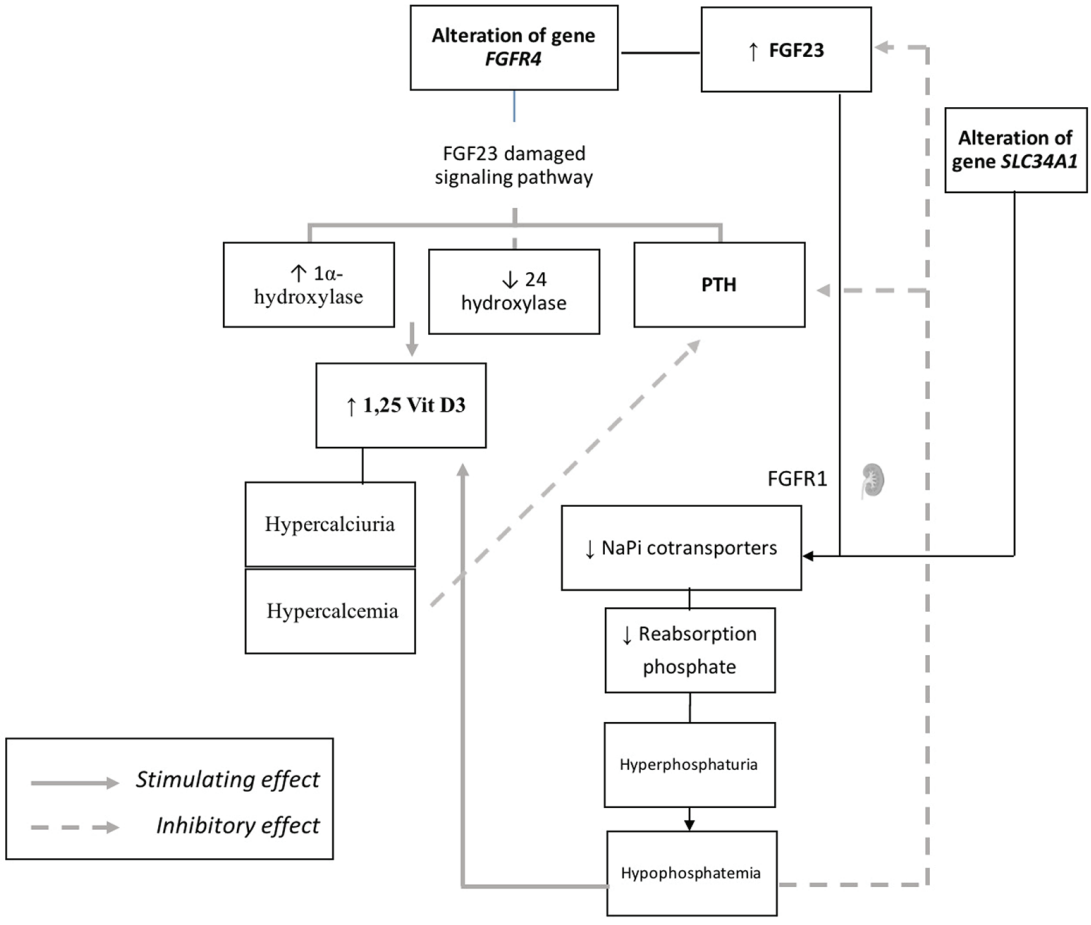

Figure 3. The damaged signaling pathway due to an alteration of gene *FGFR4* may cause an activation of PTH and 1,25 $(OH)_2$ vitamin D3, due to stimulus of the enzyme 1α hydroxylase and the lowered expression of 24 hydroxylase, thus favoring hypercalcemia and hypercalciuria. In addition, this alteration of *FGFR4* may also contribute to the inappropriately normal FGF23 level which, through binding to FGFR1, would decrease tubular reabsorption of phosphorus into the proximal tubule, exacerbating the hypophosphatemia associated with renal loss due to the inactivation of the NaPi-IIa carrier. Hypophosphatemia would be another major stimulus for increased 1,25 $(OH)_2$ vitamin D_3, although with a negative impact on the production of FGF23 and PTH

PTH: parathyroid hormone, 1,25$(OH)_2$ vit D_3: 1,25-dihydroxyvitamin D_3, FGFR: fibroblast growth factor receptor

The literature contains very few cases of nephrocalcinosis in Sotos syndrome patients. Saugier-Veber et al. (5), 2007, published three cases due to deletion with associated nephrocalcinosis, suggesting a genetic predisposition in the deletion area. Kenny et al. (6), 2011, presented two pediatric cases of 5q35 chromosome microdeletion that included *NSD1* and *SLC34A1*, explaining the wider phenotypic spectrum. This last gene encodes an important renal phosphate carrier (NaPi-IIa) and mutations thereof have been associated with nephrolithiasis/osteoporosis, hypophosphatemia 1 (OMIM #612286), Fanconi reno-tubular syndrome 2 (OMIM #613388) and Hypercalcemia infantile 2 (OMIM #616963). Recessive mutations of this gene have been associated with Fanconi syndrome and hypophosphatemic rickets, as well as familial cases of hypophosphatemia and nephrocalcinosis (7,8). However, heterozygous mutations of *SLC34A1* in patients with nephrolithiasis and osteoporosis have been published (9,10). Moreover, Schlingmann et al. (11) described a homozygous mutation in the same gene as a cause of idiopathic hypercalcemia in familial and sporadic cases, explaining the hypercalcemia with the suppression of FGF23 caused by hypophosphatemia, caused in turn by inactivation of the renal phosphate carrier NaPi-IIa. These authors suggested that supplementation with oral phosphate could help correct calcium metabolism in patients with *SLC34A1* mutation, although this treatment was not tolerated in our patient.

More recently, overlapping phenotypes associated with *SLC34A1*, *SLC34A3* and *CYP24A1* mutations have been described, and that not all the patients showed improvements in hypercalciuria and nephrocalcinosis, despite improvement in hypercalcemia and 1,25 $(OH)_2$

vitamin D_3 levels, as happened in the current case report (12,13). Moreover, an attenuation of renal phosphate wasting with advancing age has been observed, which may reflect the decreasing importance of NaPi-IIa for phosphate homeostasis over time, and other studies found impaired kidney function at a mean age of 23.8 years, even in subjects with a heterozygous mutation, suggesting the need for long-term follow-up of these patients (14,15).

Mutsaers et al. (16) described a case of Sotos syndrome due to 5q35 microdeletion that affected *NSD1* and the FGF receptor gene 4 (*FGFR4*), presenting with transient hypercalcemia but without nephrocalcinosis, in contrast to the present case report. It was suggested that the case reported by Mutsaers et al. (16) implied the existence of a change in the expression of FGF receptors (FGFR) during human renal development and that the expression of FGFR4 decreased with age. The authors proposed that the heterozygous microdeletion detected caused inactivation of this FGF23 receptor, leading to damaged signaling at an early stage of development, thus affecting calcium homeostasis, mainly mediated by FGF23 binding to FGFR3 and FGFR4. Under normal conditions, osteocytes increase FGF23 release in response to elevated calcemia, This mechanism is aimed at achieving a negative calcium balance through decreasing 1,25 $(OH)_2$ vitamin D_3 and levels of PTH, which cannot be achieved when this signaling pathway is damaged.

Considering the complex regulation mechanisms of mineral metabolism and taking into account the studies published and the findings in our case, which showed normal intact *FGF23* values, we suggest that the damaged signaling pathway caused by the alteration to *FGFR4* may have activated 1,25 $(OH)_2$ vitamin D_3, thus favoring the development of hypercalcemia and hypercalciuria. In addition, this alteration of *FGFR4* may also contribute to the inappropriately normal FGF23 level, which may in turn inhibit the expression of renal carriers NaPi of the proximal tubule through binding mainly to FGFR1. This would constitute an additional stimulus for the renal loss of phosphate and hypophosphatemia in the presented patient, already boosted by the inactivation of carrier NaPi-IIa due to the mutation of *SLC34A1*. Although hypophosphatemia was not very significant, due to the heterozygous mutation of gene, and that this defect could be partially compensated by the NaPi-IIc contransporter, it may have been another major stimulus for the increase of 1,25 $(OH)_2$ vitamin D_3 (17,18). On the other hand, the normalization of cystatin C, NAG values and renal water handling suggested an association with neonatal kidney injury and treatment with aminoglycoside (Figure 3).

Lastly, the patient's growth decreased in the most recent period after an initial period of overgrowth. This may be explained by the alteration of several genes, as delayed growth in hypophosphatemic syndromes is common, although not constant and variable. Furthermore, the regulatory action of FGF23 on bone mineralization widely recognized (19,20).

Conclusion

In conclusion, nephrocalcinosis is a phenotypic characteristic that has been reported in cases of Sotos syndrome due to deletions that include *SLC34A* and other genes associated with phosphate wasting, hypercalcemia, hypercalciuria and elevated 1,25 $(OH)_2$ vitamin D_3 levels. In these patients, long-term follow-up is recommended due to the risk of impaired kidney function, although the optimal treatment for affected patients is unknown and would require much more evidence, including genotype-phenotype relationships for different combinations of variant genes.

Ethics

Informed Consent: Consent form was filled out by all participants.

Footnotes

Authorship Contributions

Surgical and Medical Practices: Juan D. González-Rodríguez, Esther Q. Inglés-Torres, José E. Cabrera-Sevilla, Salvador Ibáñez-Micó, Francisca Bermejo-Costa, Ascensión Vera-Carbonell, Juan A. Bafalliu-Vidal, Pedro Cortés-Mora, Ana Lorente-Nicolás, José María Donate-Legaz, Concept: Juan D. González-Rodríguez, Esther Q. Inglés-Torres, Design: Juan D. González-Rodríguez, Esther Q. Inglés-Torres, José E. Cabrera-Sevilla, Salvador Ibáñez-Micó, Francisca Bermejo-Costa, Ascensión Vera-Carbonell, Juan A. Bafalliu-Vidal, Pedro Cortés-Mora, Ana Lorente-Nicolás, José María Donate-Legaz, Data Collection or Processing: Juan D. González-Rodríguez, Esther Q. Inglés-Torres, Analysis or Interpretation: Juan D. González-Rodríguez, Esther Q. Inglés-Torres, José E. Cabrera-Sevilla, Salvador Ibáñez-Micó, Francisca Bermejo-Costa, Ascensión Vera-Carbonell, Juan A. Bafalliu-Vidal, Pedro Cortés-Mora, Ana Lorente-Nicolás, José María Donate-Legaz, Literature Search: Juan D. González-Rodríguez, Esther Q. Inglés-Torres, Writing: Juan D. González-Rodríguez, Esther Q. Inglés-Torres, José E. Cabrera-Sevilla, Salvador Ibáñez-Micó, Francisca Bermejo-Costa, Ascensión Vera-Carbonell, Juan A. Bafalliu-Vidal, Pedro Cortés-Mora, Ana Lorente-Nicolás, José María Donate-Legaz.

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