Successful treatment of severe hypercalcemia in an infant with Williams syndrome using a single infusion of pamidronate followed by low calcium diet

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BACKGROUND

Williams-Beuren syndrome (WBS) is a rare genetic disorder caused by the deletion of 26-28 genes on chromosome 7. Fifteen percent of WBS patients present with hypercalcaemia during infancy, which is generally mild and resolves spontaneously before the age of 4 years. There are, however, reported cases with severe hypercalcemia that did not respond to traditional therapy and pamidronate proved to be a successful treatment in these cases . In our case with WBS and severe hypercalcemia, a single IV pamidronate infusion was successful in the treatment of the severe hypercalcemia.We also stress on the importance of a low calcium diet to further stabilize serum calcium levels in these patients.

METHODS

The patient was a 9 month old female who had not been previously diagnosed to have any illness. She presented with a 2 month history of decreased activity, poor appetite, polyuria, polydipsia and failure to gain weight. She is the first born baby to a consanguineous couple and was a product of a full term normal vaginal delivery with a birth weight of 2.85 kg. She was admitted to the NICU due to hyperbilirubinemia for 10 days. She was on a regular infant formula and was not on vitamin D supplement. On examination, she had stable vital signs but had moderate signs of dehydration with dry mucous membranes and sunken eyes. She had the typical facial features of WBS (figure 1). Other system exam was normal including the cardiovascular system. She was polyuric. Her laboratory results are shown in table 1. It was significant for severe hypercalcemia. Renal ultrasound showed nephrocalcinosis.Williams-Beuren syndrome was confirmed by FISH technique. Treatment was started with the traditional intravenous hydration and frusemide but hypercalcemia persisted. A single Pamidronate infusion with a dose of 1mg/kg was administered in an attempt to treat the hypercalcemia. No significant side effects were noted.

Figure 1.
Photo of the patient (with permission) showing classical facial features of WBS; elfin facies,depressed nasal bridge, long philtrum and small jaw.

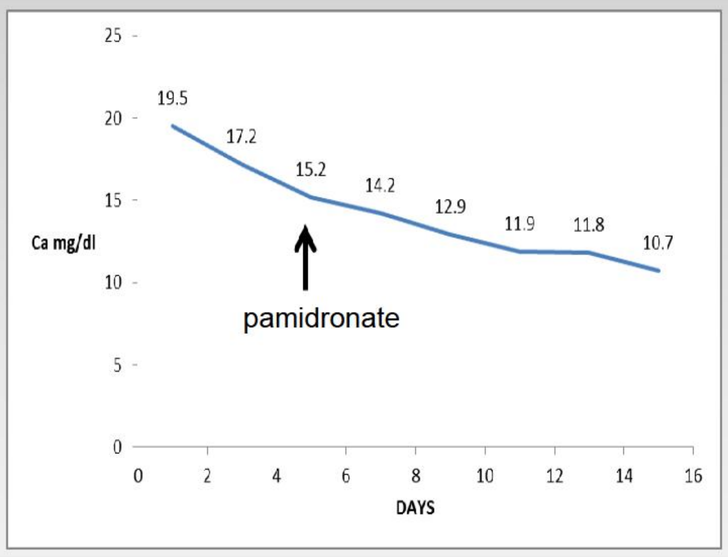
	value	normal
HB	11.3 g/dl	10.5-13
Glucose	92 mg/dl	70-100
Na	141 mmol/L	135-145
k	4.7 mmol/L	3.5-5
Ca	19.5 mg/dl	8.5-10.5
Mg	2.2 mg/dl	1.58-2.55
Ph	5.6 mg/dl	4.5-6.5
ALP	104 U/L	<450
PTH	3.4 pg/ml	14-68
Vitamin D	56.3 ng/ml	30-60
Urea	43.6 mg/dl	15-46
Creatinine	0.43 mg/dl	0.2-0.4

Table 1.
Lab results on admission

RESULTS

After the intravenous infusion of pamidronate, calcium levels declined gradually as shown in figure 2. However, a low calcium diet and a low calcium formula were necessary to keep the serum calcium level in the normal upper range. This is still needed till now, almost one year after the infusion. Her polyuria resolved and she is gaining weight.

Figure 2.
Ca serum levels before and after pamidronate infusion



CONCLUSIONS

1.Hypercalcemia associated with Williams-Beuren syndrome can be very severe with significant morbidity.
2. Pamidronate intravenous infusion proves to be effective in the treatment of this condition.
3. Low calcium diet is essential, in certain cases, to prevent the relapse of hypercalcemia associated with WBS.

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