**CASE REPORT**

**Sibling 1**
11 year-old, girl, admitted for difficulty in walking, lomber pain and inability to run  
**History:**  
- Operated for bilateral congenital cataracts – 1.5 years-old  
- Retarded growth and development – 2.5 years-old  
- Retinal detachment – 10 years-old  
- Parents are third degree cousins

**Physical examination:**
- Weight: 33.2 kg (-0.65 SDS), Height: 125.6 cm (-2.6 SDS)  
- Disproportionate short stature (Upper/Lower ratio 0.76)  
- Pectus excavatum deformity, increased lomber lordosis and scoliosis  
- Horizontal nystagmus and mild mental retardation

**Laboratory tests:**
- Ca, P, ALP were within the normal range  
- 25-OH vitamin D: 16.23 mcg/L

**Sibling 2**
7 year-old boy was unable to walk.
**History:**
- Bilateral congenital cataracts  
- Severe mental retardation  
- Global motor and developmental delay

**Physical examination:**
- Weight: 25 kg (0.49 SDS), Height: 108 cm (-2.63 SDS)  
- Proportionate short stature (upper/lower ratio: 0.96)  
- Kyphosis, increased lumbar lordosis

**Laboratory tests:**
- Ca, P, ALP were within the normal range  
- 25-OH vitamin D: 6.5 mcg/L

The same homozygote mutation was detected in patient’s seven year old brother via family screening.

![Image 1](https://example.com/image1)

**Figure 1:** (a) Physical features of the patient at admission, X-rays of the patient at (b) admission and (c) following the first year of bisphosphonate treatment.

Spondyloocular syndrome  
**XYLT2** sequence analysis  
→ homozygous mutation on  
11. exome c.2548G>A (p.Aspl850Asn)

![Image 2](https://example.com/image2)

**Figure 2:** (a) Physical features of the patient’s brother at admission, X-rays at (b) admission and (c) following the first year of bisphosphonate treatment.

- 1 mg/kg bisphosphonate ( pamidronate) treatment every three months for a year  
- Vitamin-D replacement

L1-L4 bone mineral density z-scores improved in both siblings.

**CONCLUSION**

Treatment options for osteoposis in spondyloocular syndrome are scarce and data on results of bisphosphonate therapy are limited. The increase in BMD z-score suggests that bisphosphonate treatment can be beneficial for osteoporosis in patients with spondyloocular syndrome. More data is required to understand long term effects of bisphosphonate treatment for osteoporosis in this syndrome.