

following 12 months despite brace treatment. Four months after the initial hip dislocation, left calcaneal fracture occurred of unrecognized cause. An 11-year-old boy developed separation of the left proximal tibial physis after very minor trauma during brace treatment for the Charcot joint in the contralateral knee.

Discussion

Because of their insensitivity to superficial and deep painful stimulation, which is sometimes accompanied by intellectual disability and self-mutilating behavior, patients with CIPA may overuse their extremities, leading to fractures and dislocations. Although skeletal-system complications are more common in the lower limbs, almost all bones and joints may be affected. Szöke et al. [14] reported that 58 % of patients with CIPA had osteoarticular disorders of the feet and ankles, 53 % of the knees, and 26 % of the hips. Bar-On et al. [15] reported that in 31 fractures in patients with congenital insensitivity to pain, 28 occurred in the lower limbs. These findings that bone disorders occur more frequently in the lower limbs are consistent with ours. Compared with our PRG patients, on average, patients in the LRG had fewer skeletal complications, probably because the patients attending our institution were older than those identified by the literature review. In addition, most patients came to our institute for management of skeletal problems.

Fractures occurred in the early years of life, especially between the ages of 4 and 6 years. Fractures occur frequently at these ages because young children are very active. As children get older and become less active, the frequency of fractures decreases. However, in our study, joint dislocations and infections had no apparent relationship with age. Previous studies have not documented the age of onset of bone disorders in CIPA and similar conditions [14, 15].

The causes of most fractures and dislocations were not documented. Where they were, some were caused by minor trauma, such as short falls. This suggests that CIPA patients may sustain injuries from minor traumas that would not result in injuries in normal children. Video gait analysis of young CIPA patients indicated that they walk fast and their heels do not decelerate smoothly before foot contact, which may contribute to the high incidence of injuries in the lower limbs [16]. In our study, two patients in PRG experienced multiple and/or repeated musculoskeletal conditions during the follow-up period, and the second injury occurred with unrecognized cause or after very minor trauma. When treating some musculoskeletal conditions, the medical staff must keep in mind the possibility of additional or repeated injury.

Conservative therapy was used more frequently than surgery to manage fractures, joint dislocations, and Charcot joints, but surgery was more used to manage infections. Köster et al. [17] reported that surgical improvement of the anatomy may not prevent progressive joint dislocation in CIPA patients. Furthermore, cardiovascular complications such as bradycardia, and hypotension following anesthesia, are common in patients with CIPA [18]. Therefore, surgical treatment of these patients may not always be advisable. On the other hand, use of casts, a representative conservative treatment, has drawbacks. First, the intellectual disability and loss of pain sensation of CIPA patients promotes instability. Second, there is a risk of developing pressure sores because of the sensory disturbance. In our study, the frequency of complications with surgical management was higher than that with conservative management, though the actual frequency may be higher in both managements because most of results came from the literature review.

To sum up, because both surgical and conservative treatments have their drawbacks, preventing trauma is very important in this patient group [19]. While very few comments about injury prevention appear in the medical literature, some of our PRG patients wore high-top sneakers and knee pads, and the parents covered the floor with sheets of soft material to prevent injuries. In addition, a few patients even used wheelchairs for locomotion to prevent accidental falls despite their walking ability.

One limitation of this study is that all LRG patients were drawn from retrospective studies. Because CIPA is an extremely rare disease, collecting information on many patients is difficult. We therefore combined LRG and PRG data, which made analysis of 91 patients possible—the largest group reported to date. Though diagnoses of only 9/91 patients were confirmed by gene analysis, we judged that clinical findings, physiological tests, and, in some cases, nerve biopsy led to the correct diagnosis in the other patients.

In conclusion, most Japanese patients with CIPA have skeletal complications, most of which occur in the lower limbs. Fractures are frequent between 1 and 7 years of age, whereas other bone disorders are apparently unrelated to age. The major known causes of bone disorders in this study were minor trauma such as short falls. Conservative therapy was used more frequently than surgery to manage fractures, dislocations, and Charcot joints.

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Conflict of interest None.

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