



## Correspondence

## Chondrodysplasia Punctata of Hip Joints on Routine Radiography Provided a Diagnostic Clue of Zellweger Syndrome

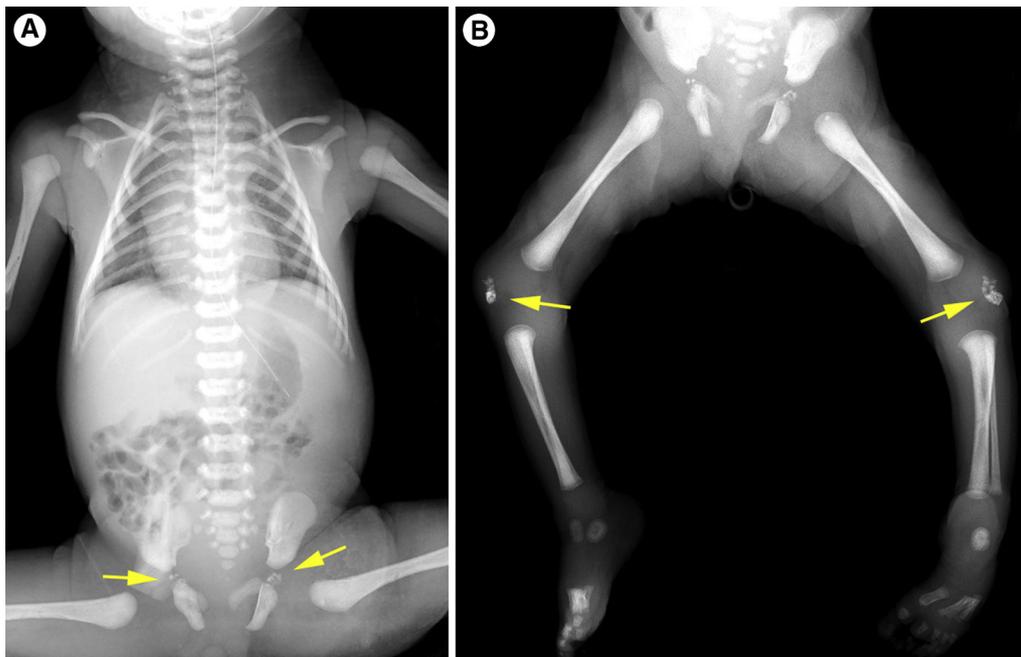


## To the Editor:

We read with great interest the report by Bamford.<sup>1</sup> He emphasized that chondrodysplasia punctata (CDP) was a cardinal sign of Zellweger spectrum disorders and that routine radiography should be carefully screened. We wish to report newborn with CDP of the hip joints in whom routine radiography led to an early diagnosis of Zellweger syndrome.

This one-day-old term female, with a birth weight 2046 g, was referred to the neonatal intensive care unit with hypotonia and

respiratory failure. A large anterior fontanel with a prominent forehead was noted during the examination. Radiography of the abdomen and extremities revealed CDP of both the hip joints and patellae (Fig A, B). Serum aspartate transaminase was elevated to 804 U/L at age 15 days. Subsequently, we confirmed an elevated level of very-long-chain fatty acids (C24:0, C25:0, C26:0), which was suggestive of peroxisomal disorders. Compound heterozygous mutation in *PEX1* was identified, consistent with Zellweger syndrome. Owing to poor prognosis and lack of significant treatment,<sup>2</sup> we initiated symptomatic treatment with chenodeoxycholic acid



**FIGURE.** (A) Routine radiography of the neonate shows calcific stippling in both hip joints symmetrically (arrows). Because peroxisomal diseases were highly suspected, we conducted bone radiography of the extremities. (B) On the bone radiography of extremities, calcific stippling in both the patellae was indicated (arrows), which was reported as a cardinal sign of Zellweger syndrome. The color version of this figure is available in the online edition.

and vitamin K for bile acid abnormality and phenobarbital, levetiracetam, and carbamazepine for the partial seizures. At age six months, the patient was discharged from the hospital and received supportive care at home.

Because Zellweger syndrome is often confused with other diseases presenting with hypotonia, the importance of CDP of the patella has been emphasized in recent years.<sup>3,4</sup> Early diagnosis in newborns is important for prolonging the time spent with their families.<sup>5</sup> This patient demonstrates that routine radiography can provide a critical diagnostic clue and lead to the early diagnosis of Zellweger syndrome.

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## References

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## Correspondence Reply to Kitaoka et al.



To the Editor:

I wish to reply to Kitaoka et al.<sup>1</sup> I thank the authors for citing my recent Clinical Letter in *Pediatric Neurology* entitled “Chondrodysplasia punctata: A clue to the Zellweger spectrum disorders.” In the report, I presented an interesting and perhaps unique patient in whom Zellweger syndrome was accompanied by chondrodysplasia punctata (CDP). Although it remains unclear if CDP is a cardinal sign of all Zellweger spectrum disorders, it is important to know that another patient with Zellweger syndrome manifested with CDP. This second case again illustrates that routine radiography should be carefully screened in suspected cases.

## Reference

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