

Living with a rare disease, the diagnosis is the key

The awareness of rare conditions in general practice is essential to optimize children's fair chances in society around the world. Failure to thrive, recurrent infections, skeletal abnormalities are signs of Shwachman Diamond Syndrome. An early accurate diagnosis leads to proper treatment, reduction of symptoms and avoids life threatening complications.

In many middle and lower economies there is little access to efficient and affordable diagnostics among providers of child health. The appeal to international awareness of rare disease must concord with actions to adopt diagnostic facilities in a widespread practice.

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