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Answers to Medical Quiz

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A1. Diagnosis: Achondroplasia
The diagnosis is suggested by the following physical finding: Disproportionately large head, prominent forehead, depressed nasal bridge, rhizomelic micromelia, short stature and lordosis. Increased pelvic tilt producing protuberant abdomen and prominent buttocks. Trident hand with short phalanges. The condition can be detected by birth. Gene location: 4p16.

A2. Skeletal Survey. The x-ray findings are pathognomonic for achondroplasia at any age.

A3. Differential diagnosis: Diabetic embryopathy (maternal diabetes).

A4. Associated findings and complication: The condition can be benign with no impact on life span. Some patients may suffer from recurrent otitis media, conductive or sensorinural deafness and possible psychological problems related to body image.

A5. Risk of recurrence to patient's children: The condition is Autosomal Dominant. There is 50% risk for patients' children to be affected. This case is due to fresh mutation as the mother and father are normal. Genetic counseling is recommended.

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