Hypochondroplasia (H.) has been considered for a long time a mild form of achondroplasia (A.). Both entities, on the contrary, are definitely two separate conditions, hypochondroplastic parents having never had children with A. and vice versa. H. however may exhibit clinical and radiographic variations, so that in a severe form, it may resemble A. H. is not a rare hereditary chondrodystrophy, although relatively few cases have been reported in the literature. This probably depends on the numerous though mild clinical and radiological features, which suffer from a lack of specificity and are also seen in other types of disproportionate dwarfism.

Some patients may be almost normal radiologically and the diagnosis can be made only on the findings of short stature with a relatively long trunk and disproportionate short limbs. Consequently, mild cases may easily be missed and erroneously labelled as constitutionally short stature. There are likely to be many individuals in the population with unidentified H. The disorder is of autosomal dominant inheritance, even though most individuals present themselves as new mutations. For these subjects the elevated paternal age is considered an effective element, according with the known genetic increased risk of gene mutation with older paternal age.

Hypochondroplastic patients are generally almost normal in appearance except for short stature and disproportionately short limbs compared with the trunk. The range of final height is between 127 and 152 cm. However, one physical feature that tends to make examiners consider them abnormal in appearance is macrocephaly. For a long time, macrocephaly has been considered as an occasional manifestation of H.

In numerous series, however, the increased skull size was already noted at birth in more than 50% of the cases. The data disagree with the findings of normocephalic dwarfism previously stated. Lumbar lordosis and genu varum are frequently noted. On the contrary, mental retardation and limited elbow extension are less frequent. The age of manifestation is school-age with full-blown signs. Diagnosis can rarely be made at birth.
At this time, in the absence of more specific knowledge of the origin and the nature of the underlying biological abnormality, radiological examination alone can establish a correct diagnosis.

Hall et al. (2) propose the following primary criteria as very frequently present for the radiological diagnosis of H.:

1. The lumbar interpediculate distance is narrowed or unchanged. This means altered, owing to the normally increased distance from above downward from L1 to L5 (Fig. 1a).
2. The iliac bones are squared and shortened, with broad lower portions. The sciatic notches are small (Fig. 1b).
3. The femoral necks are short and broad (Fig. 1b).
4. The tubular bones are shortened and relatively squared and the metaphyses are wide. The long bones, generally speaking, appear plump. The shortening of the limbs is without obvious rhizomelic, mesomelic or acromelic predominance.
5. There is generalized brachydactyly, even if in a few cases the hand can be absolutely normal (Fig. 1c).

The metacarpophalangeal pattern profile shows equal and balanced shortening of all bones. Other radiological signs less frequent than the above, and which can be evaluated as secondary criteria, are the following:

A. The lumbar pedicles are shortened so that the spinal canal seems less deep. Such "spinal canal stenosis" rarely causes neurologic complications; if they occur they are very mild, such as occa-