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特許

1）特許権（発明者）

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Ⅴ．診断指針

低フォスファターゼ症の診断指針

主症状

1. 骨石灰化障害
   骨単純X線所見として骨の低石灰化、長管骨の変形、くる病様の骨幹端不整像
2. 乳歯の早期脱落(4歳未満の脱落)

主検査所見

1. 血清アルカリフォスファターゼ(ALP)値が低い(年齢別の正常値に注意)

参考症状

1. ビタミンB6依存性けいれん
2. 四肢短縮、変形

参考検査所見

1. 尿中フォスフォエタノールアミンの上昇(尿中アミノ酸分析の項目にあり)
2. 血清ピロリン酸値の上昇
3. 乳児における高カルシウム血症

遺伝子検査

1. 確定診断、病型診断のために組織非特異的ALP (*TNSALP*)遺伝子検査を行う事が望ましい

参考所見

1. 家族歴
2. 両親の血清ALP値の低下

診断基準

主症状一つ以上と血清 ALP 値低値があれば遺伝子検査を行う。参考症状、参考検査所見、参考所見があれば、より確実である。

症例の御相談、遺伝子検査の御依頼、血清ピロリン酸測定の御依頼は、厚生労働省難治疾患克服事業「低フォスファターゼ症」研究班事務局(hypophos@ped.med.osaka-u.ac.jp)までお問い合わせください。