Hypertrichosis, gingival hypertrophia, epilepsy and mental retardation: a separate entity?

This syndrome was maybe described for the first time with the famous Julia Pastrana [1834-1860] who presented congenital hypertrichosis terminalis , gingival hyperplasia and maybe a mental retardation as described by waterman “feeble intellect” (1) though others (Laurence 1857 , Sigmund (1) found her “ intelligent”) ; however, no form of epilepsy was reported. Some authors (Snyder (2), Anavi et al. (3), Kiss et al. (4)) have noticed that quadruple associations in some rare cases.

We report a new case of this syndrome.

Case report

A 7 years old Caucasian boy, product of healthy non consanguineous parents, presented a general hypertrichosis, gingival hypertrophia, a mental retardation and epilepsy. He has no familiar pathological history. The pregnancy was unremarkable, delivery at term uneventful. The mother didn’t report any medication intake. On examination he had thick eyebrows and generalized hypertrichosis with a promaxilly, a flattened nose, low-set ears, and a large cephalo-conchal angle (Figures 1 - 2). On oral examination (Figure 3) we found a severe gingival hypertrophia which covered almost the whole

Figure 1: (A) (B) Preoperative photograph of patient with thick eyebrows, a flattened nose, low-set ears and a large cephalo-conchal angle.

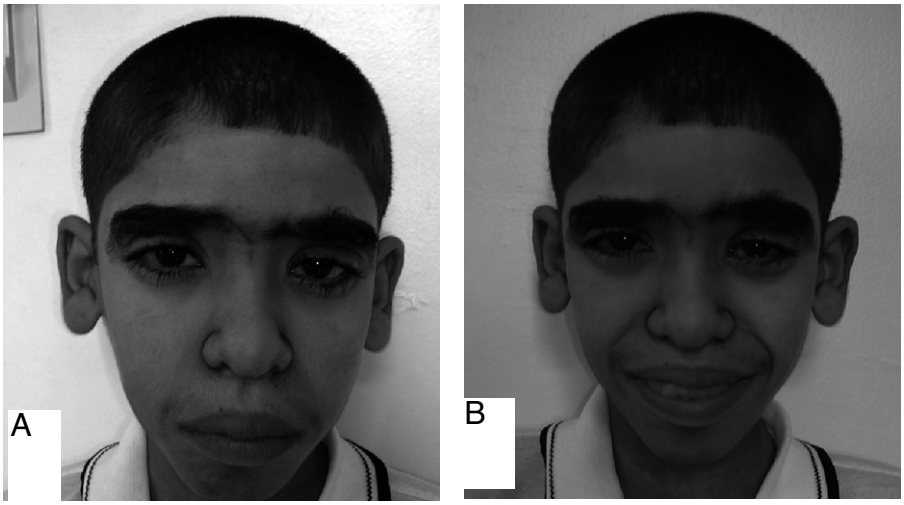


Figure 2: (C) (D) Generalized hypertrichosis

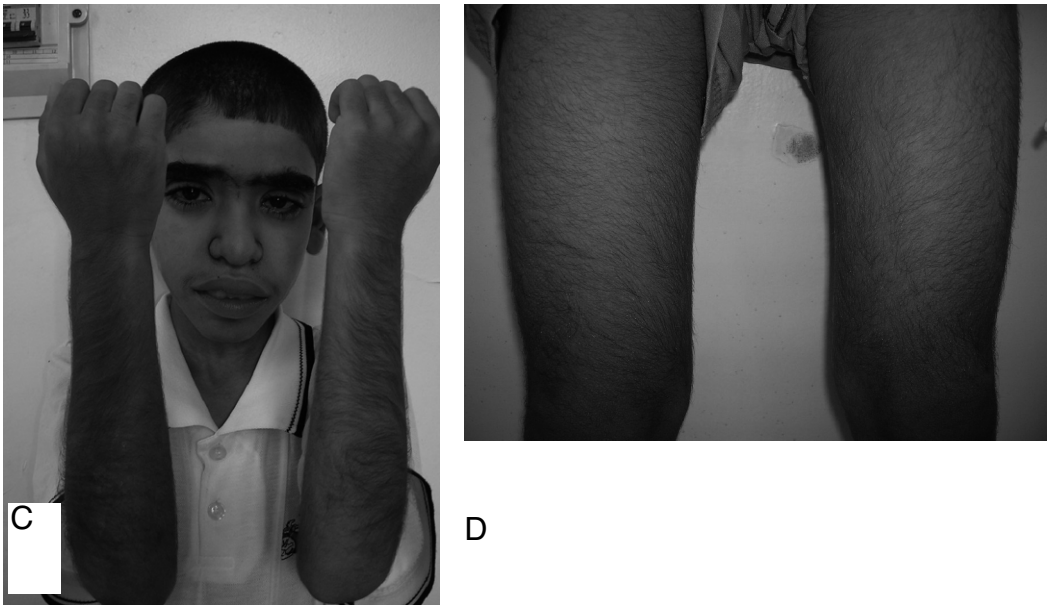


Figure 3: (E) Promaxilly with gingival smile; (F) On oral examination we found a severe gingival hypertrophia which covered almost the whole tooth. We also discovered a delayed tooth eruption in the lower arch.

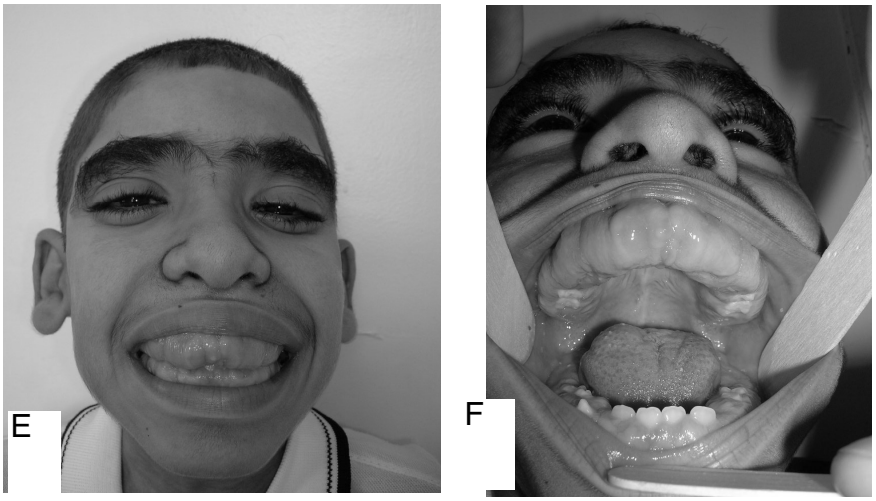
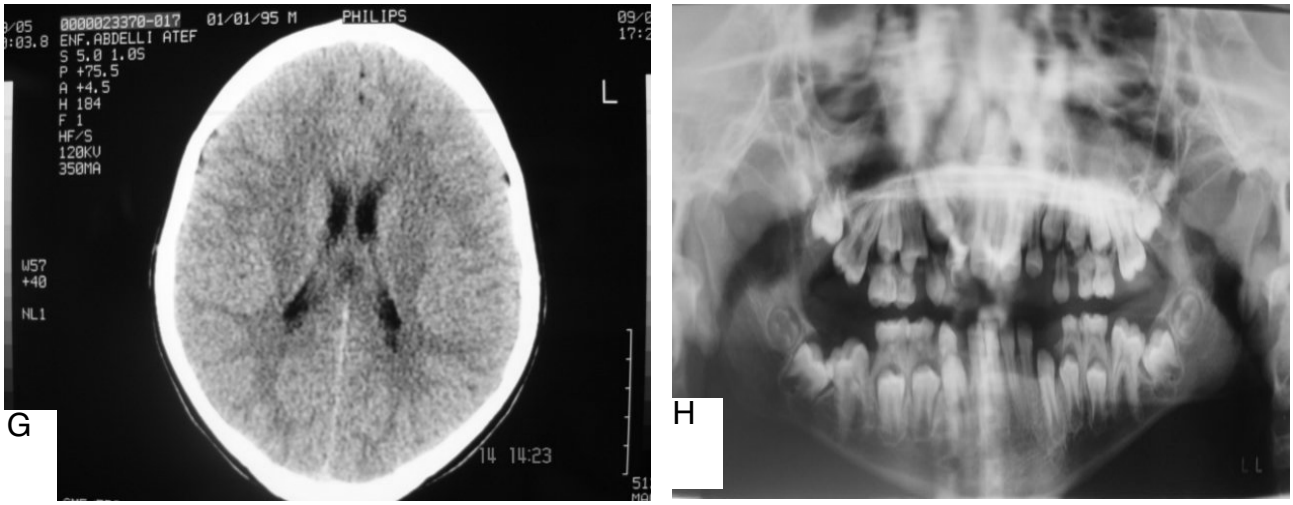


Figure 4: (G) CT scan there were no abnormalities (H) orthopantography: all teeth were present and in good position. Tooth germs are in place

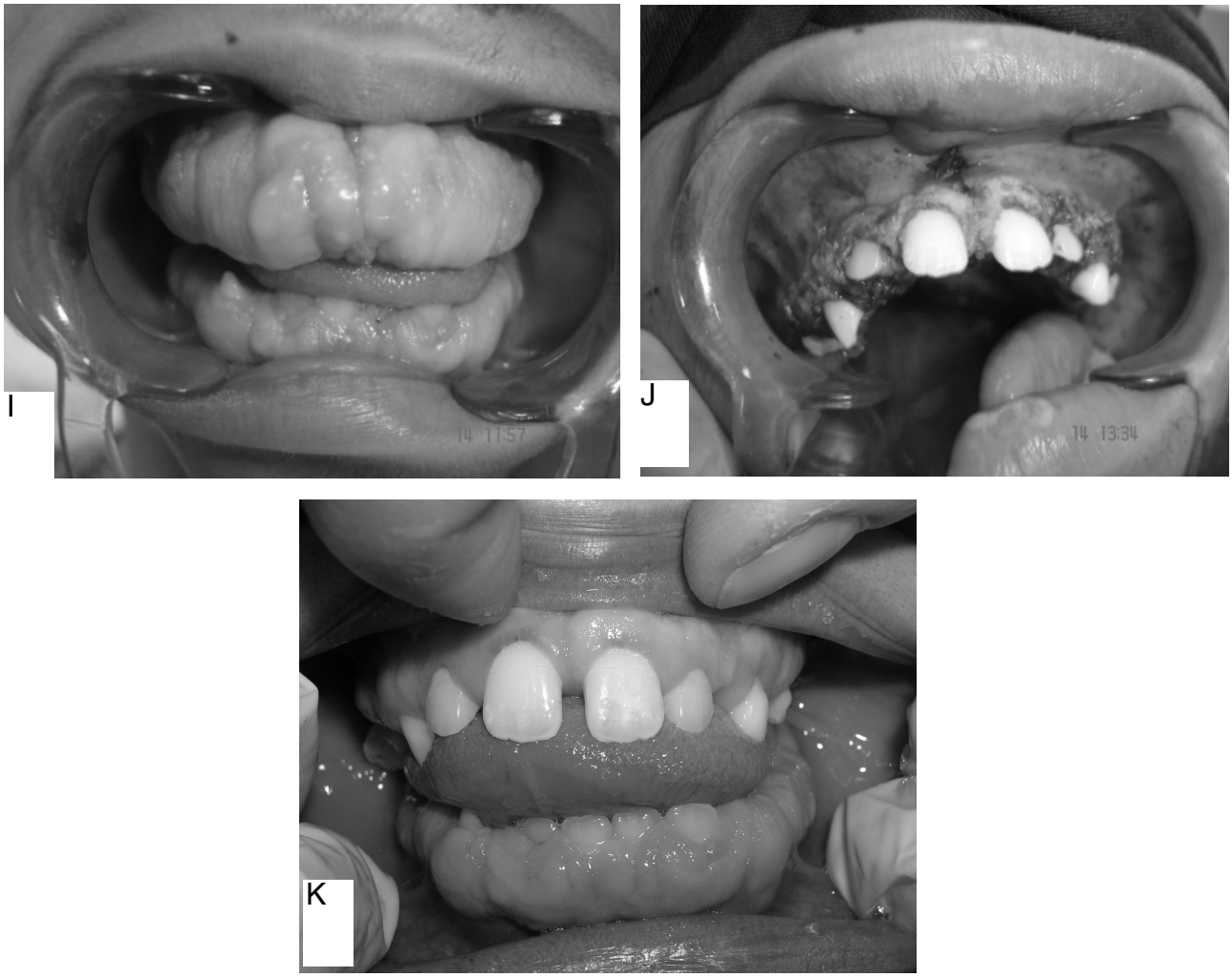


tooth. We also discovered a delayed tooth eruption in the lower arch. His cephalic perimeter was normal as well as his staturo-ponderal development. His external genitalia were normal. Notice, the first seizures occured at the age of five years. On scan CT there were no abnormalities (Figure 4-G). On orthopantographie: (Figure 5-H) all teeth were present and in good position. Tooth germs are in place. Genetic exploration revealed a normal karyotype. He underwent a gingivectomy in two stages. The evolution was good without recurrence with a decline of 4 years (Figure 6). Histological examination showed a regular squamous lining, very hyperplastic, parakeratotic and acanthotic; under which there is a dense collagenous fibrous tissue with a slight inflammatory lymphoplasmacytic infiltration.

Conclusion

Hypertrichosis and gingival hypertrophy were reported in more than fifty cases. Against their, simultaneous association with epilepsy and mental retardation has been found only in some cases. The management of this association should be multidisciplinary; and genetic exploration is certainly the key to this puzzle.

Figure 5: (I) preoperative view; (J) immediate postoperative view after gingiectomy; (K) appearance 4 years after gingivectomy with good result and without recurrence



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Adénocarcinome gastrique développé après un lymphome

L'occurrence de deux tumeurs d'histologie différente dans le même organe est extrêmement rare. Nous rapportons un cas rare de tumeur métachrone. Il s'agit d'un adénocarcinome gastrique développé 10 ans après un lymphome de l'estomac.

Observation

Il s'agissait d'un homme de 63 ans qui a été traité en 1993 pour lymphome non hodgkinien à grandes cellules de haut grade siègent au niveau de la grande courbure gastrique, traité par chimiothérapie à base de six cures de CHOP (Adriamycine, Endoxan, Oncovin, Prednisone).