EEC Syndrome (Ectrodactyly, Ectodermal Dysplasia, cleft lip/palate)
Case Report

INTRODUCTION
The EEC syndrome (ectrodactyly, ectodermal dysplasia, cleft lip/palate) is a rare genodermatosis (about 200 cases reported) caused by the absence of the transcription factor p63, encoded in the chromosome 7q11. We describe the case of a child with clinical and histopathological signs of the syndrome.

CLINICAL CASE
A three year-old male from Lima-Peru, is admitted to hospital for periapical dental abscess. The patient is the youngest of three siblings (6 and 11 years old respectively, without signs of disease). The parents are from Apurimac (highlands of Peru) and have no history of disease. The patient was a product of a full-term pregnancy, birth weight of 2,850 kg. The mother (33 year-old) revealed no abnormalities in prenatal care.

Physical examination showed thin and light hair, madarosis and bilateral loss of distal third of eyebrows, low-set ears, atrophic remnant by the absence of both nipples. Also presents diffuse xerosis cutis, café au lait spots in the anterior chest and patches of hypopigmentation following Blaschko's lines in trunk and limbs. Has a scar of a cleft lip correction surgery, hypoplastic conical teeth, with the presence of diastema. On his feet presents fusion of the first and second fingers, and also the third and fourth finger, forming ectrodactyly, in addition to nail dysplasia.

Histopathology showed thinned epidermis with no ridges and rudimentary annexes, compatible to Hypohidrotic ectodermal dysplasia.

CONCLUSIONS
The EEC syndrome is caused by the mutation of p63 gene (family of P53 tumor supressor gene) that is related to the development of limbs, epithelia and craniofacial region. Ectodermal dysplasia is manifested by alteration of two or more ectodermal structures (nail, sweat glands, hair or teeth). Because of its phenotypic variability, familial cases may occur uncomplete. Sporadic cases occur in a more severe form and with more expressivity of clinical signs, as our case report.

REFERENCES