Imaging Keratitis-Icthyosis-Deafness (KID) syndrome with FDG-PET (F18-fluorodeoxiglucose-Positron Emission Tomography)

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ABSTRACT

Keratitis-Icthyosis-Deafness (KID) syndrome is a rare dysplasia characterized by vascularizing keratitis, congenital sensorineural hearing-loss, and progressive erythrokeratoderma. To our knowledge, this is the first KID syndrome imaged with FDG-PET in the literature. This paper is intended to help familiarize with the FDG abnormalities related to this rare entity.

CASE REPORT

INTRODUCTION

Keratitis-Icthyosis-deafness syndrome (KID syndrome) (1) is a rare ectodermal dysplasia caused by heterozygous missense mutations in the connexin-26 gene, GJB2 on chromosome 13q11-q12. It is characterized by vascularizing keratitis, profound congenital sensorineural hearing loss, and progressive erythrokeratoderma, a clinical triad that indicates a failure in development and differentiation of multiple stratifying epithelia (2,3). Squamous cell carcinoma of the skin and oral mucosa is rare but a serious complication (4) that can shorten life expectancy. We present, to our knowledge, the first case of KID syndrome imaged with FDG-PET (F18-fluorodeoxiglucose - positron emission tomography).

CASE REPORT

We present the case of a 21-year-old woman diagnosed with KID syndrome who had a whole-body FDG-PET scan (Fig. 1a) in our hospital. The patient presented with long-history of left-deafness, scalp keratitis, plantar icthyosis and recurrent squamous cell carcinoma (SCC) of the second left finger confirmed by MRI (Fig. 2). The whole-body FDG-PET scan was ordered to rule out distant metastatic disease. The scan showed the local SCC recurrence and did not show evidence of metastatic disease; but multiple other unexpected abnormalities were seen, such as heterogeneous, FDG-uptake throughout the scalp (Fig. 1 arrow-a.1) and marked, diffuse FDG uptake also was seen in the plantar skin of both feet, explored further in the discussion section. These abnormalities were difficult to characterize at the time of the scan. After review of the case, we think the abnormalities are related to her rare KID syndrome. It is our intention with this case report to discuss these findings with the lectors and to familiarize our community with these rare findings.

DISCUSSION

There is very little literature about KID syndrome and to our knowledge, no literature related to FDG-PET imaging of the syndrome prior to this paper. Since we found imaging abnormalities that we think are directly related to this entity, we thought it would be important to discuss them in this case report.

The FDG-PET scan showed expected focal, high FDG-uptake in the left hand consistent with known local SCC recurrence (Fig. 3 arrow-a.3), with a SUVmax of 3.7, and no metastases as already mentioned.

Interestingly, the scan also showed very heterogeneous, FDG-uptake throughout the scalp (Fig. 1 arrow-a.1) with an SUVmax of 1.8, which correlated with multiple minimally-enhancing soft-tissue masses seen on a recent CT (Fig. 3) and with physical exam as shown in the illustration (Fig. 1b). This was thought to be inflammatory changes probably related to the patient's known vascularizing keratitis.

There also was questionable mildly increased FDG-uptake affecting the left temporal bone and inner ear without significant anatomical correlation (Fig. 1 arrow-c.1). Findings in the temporal bone associated with this syndrome have previously been described in the literature and a possible relationship with the congenital hearing loss has been hypothesized (5).

Marked, diffuse FDG uptake also was seen in the plantar skin of both feet, with a SUVmax 3.9 (Fig. 1 arrow-a.4). This finding was of unclear significance to us and after reviewing the patient's history and discussing the case it was thought to correspond to the patient's long known history of plantar ichthyosis. This finding was unexpected since ichthyosis characterizes by 'non-inflammatory' scaling of the skin, and therefore, no accumulation of FDG-avid inflammatory cells was anticipated (6).

In our single-case experience, this syndrome shows significant abnormal findings on FDG-PET imaging that are not related to cancer and can be misleading. We consider of great importance to share our experience with the rest of the community so that they can have an opportunity to familiarize with these rare findings.

TEACHING POINT

Heterogeneous FDG uptake in soft tissue and skin abnormalities related to KID syndrome can be seen with FDG-PET imaging and should not be confused with malignancy.

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FIGURES

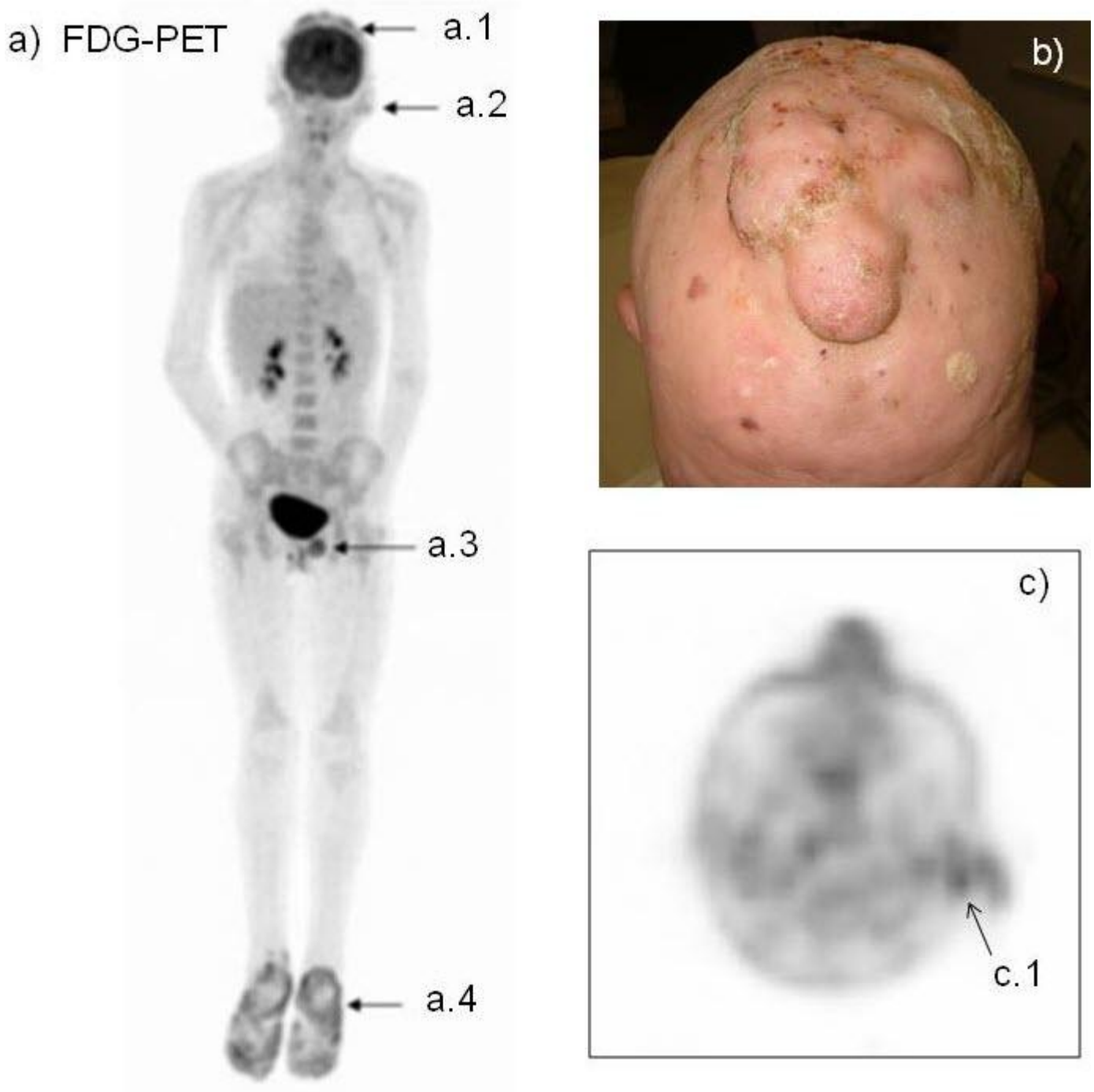

Figure 1: These images illustrate the case of a 21 year old woman diagnosed with KID syndrome. Image-a) shows the MIP (Maximum Intensity Projection) whole body FDG-PET scan of the patient after i.v. injection of 15 mCi F18-FDG. The image was acquired 60 minutes post-injection. The patient fasted for >6 hours and had a glucose level of 111 mg/dL at the time of injection. Abnormal heterogeneous FDG uptake was seen in the head and feet. However, there was no evidence of distant metastatic disease from patient's know recurrent SCC in the second finger of the right hand. The scalp showed heterogeneous, nodular, mild FDG uptake throughout the patient's extensive subgaleal soft tissue, likely inflammatory. The feet showed abnormally high, diffuse, FDG uptake through the skin of the patient's feet soles. No evidence of hypermetabolic lymphadenopathy or masses. There was no evidence of multifocal, erratic, high FDG activity pattern suspicious for metastatic disease.

Arrow-a.1 points at the heterogeneous FDG-uptake seen throughout the scalp, which correlated with multiple soft-tissue masses on physical exam as demonstrated on image-b). Arrow-a.2 points at the questionable mildly increased FDG-uptake affecting the left temporal bone and inner ear, better seen on axial cross-sectional image at that level (image-c; arrow-c.1). Arrow-a.3 points at the abnormal, focal, high FDG uptake seen in the patient's left hand, which corresponded to the patient's known recurrent SCC in the second finger (also confirmed by MRI, image not shown). Arrow-a.4 points at the marked, diffuse FDG uptake seen in the plantar regions, corresponding to the patients known ichthyosis. This finding was unexpected since ichthyosis is a skin disorder characterized by 'non-inflammatory' scaling of the skin.

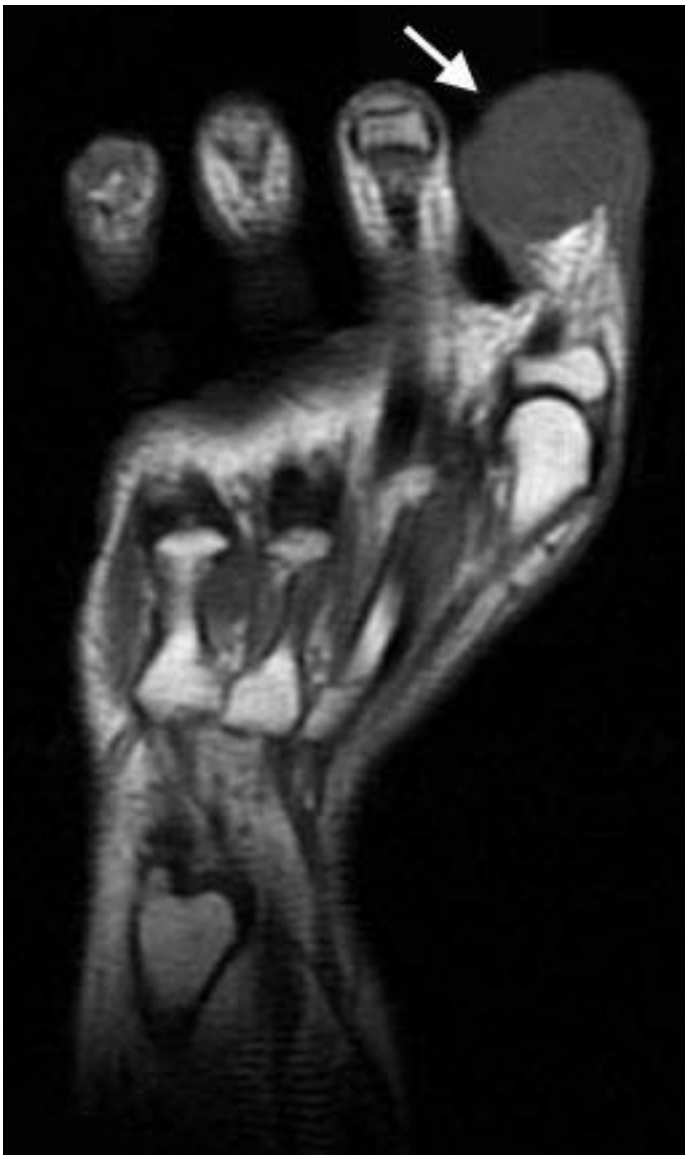

Figure 2: A 21 year old woman with KID Syndrome and a history of squamous cell carcinoma of the right index finger status post amputation. A new necrotic appearing mass is observed on the distal second digit (arrow), with the first digit out of plane. MRI Image (1.5 Tesla magnet) shows patient's left hand, captured with coronal T1-weight images with STIR sequence as fat suppression technique and coronal T1 with fat saturation post-contrast (Gd). Image shows signal abnormality in subcutaneous tissue of second phalanx and increased T2 signal and enhancement at the distal portion of the second proximal phalanx adjacent to mass.

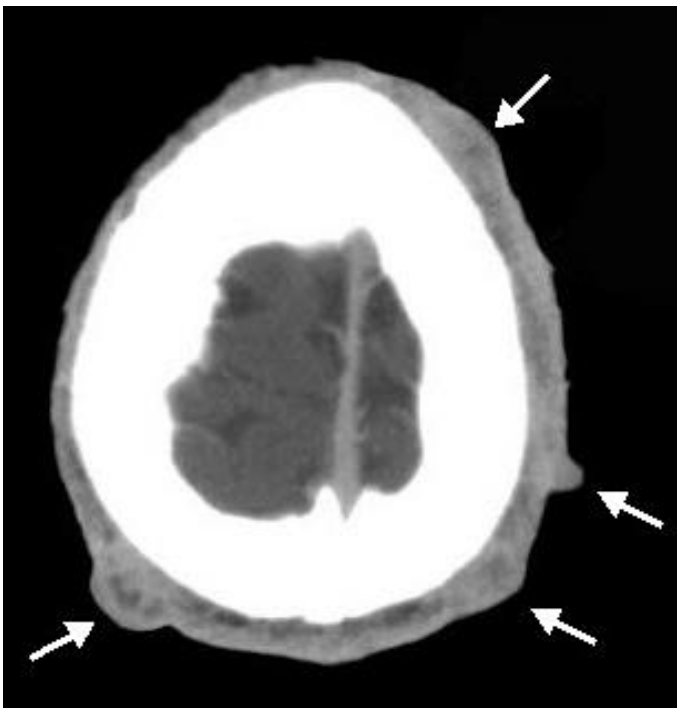

Figure 3: A 21 year old female diagnosed with KID Syndrome with a history of sqamous cell carcinoma. Image taken from multiple contiguous axial 3.75-mm images from the foramen magnum through the vertex after administration of 110cc Omnipaque-350 i.v. contrast agent, taken at standard time. Images taken with 16-slice scanner at 200 mA and 120.0 kV. Image shows extensive subgaleal soft tissue, beginning at the vertex and involving the right and left extracalvarial soft tissue extending to the posterior right parieto-occipital region (arrows).

Etiology	Heterozygous missense mutations in the connexin-26 gene, GJB2, on chromosome 13q11-q12.
Incidence	Unknown
Gender Ratio	Unknown
Age Predilection	At birth
Risk Factors	Genetic: autosomal dominant mode of inheritance
Treatment	Can only treat clinical symptoms and perform surveillance for malignancy; nothing specific for KID Syndrome.
Prognosis	Poor quality of life. Associated malignancies may result in shortened life expectancy.
Findings on Imaging	Unexpected FDG uptake in abnormal soft tissue and skin disease associated with KID Syndrome.

Table 1: Summary Table: KID Syndrome

	Hand/Finger	Skull	Feet	Whole Body
MRI	Tumor recurrence with T1 signal abnormality in subcutaneous tissue and increased T2 signal and enhancement.			
PET-FDG	Tumor recurrence	Mild inflammatory changes in amorphous soft tissue mass.	Diffusing FDG consistent with high inflammatory skin changes.	KID Syndrome
CT		Amorphous soft tissue mass with mild heterogeneous contrast enhancement.		

Table 2: Imaging Findings in KID Syndrome

ABBREVIATIONS

FDG = F18-fluorodeoxiglucose
GD = Gadolinium
KID syndrome = Keratitis-Icthyosis-deafness syndrome
MIP = Maximum Intensity Projection
PET = Positron Emission Tomography
SCC = Squamous cell carcinoma

KEYWORDS

Keratitis, Icthyosis, deafness, KID syndrome, FDG, PET