INFECTIOUS DISEASES (BACTERIAL, FUNGAL, VIRAL, PARASITIC, INFESTATIONS)

FIRST CASE OF TRICHODYSPLASIA SPINULOSA IN ITALY: CLINICAL REPORT AND VIRAL CHARACTERIZATION

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Background: Trichodysplasia spinulosa (TS) is a rare, disfiguring skin disease affecting immunocompromized patients, characterized by papules and spines on the face, alopecia of eyebrows and lashes, and thickening of the affected skin layers. It has been reported for the first time in 1999, but only in 2010 the causative member of Polyomavirus family (TSPyV) has been sequentiated. To date, up to 35 TS cases have been reported worldwide, none in Italy.

Observation: Here, we present the first Italian case of TS in a 8 year-old kidney transplant recipient who has been already transplanted twice for a congenital nephrotic syndrome caused by mutations in NPHS1 gene encoding nephrin. He developed the typical TS clinical feature with keratotic lesions, first noticed on his face, and gradually extended to the neck, back and extremities. Skin biopsy showed keratotic masses expanding from the hair follicles, with widened infundibuli and inner follicula layers containing increased cellularity of nucleated eosinophilic inner root sheat cells with trichohyalin granules. Subsequently, the patient underwent to virological analysis in order to characterize the causal virus of the disease: keratotic spines and urine samples confirmed high viral load for TSPyV DNA (6x106 copies/cell and 4x105 copies/mL, respectively). Moreover in urine, a huge number of “decoy cells” with enlarged ground-glass nuclei were observed alongside many other cells with degenerative changes in the cytoplasm and large hyaline granules very much resembling those observed in TS skin biopsy. In blood samples TSPyV DNA was also detected. Electron microscopy is underway. Regarding treatment, first the immunosoppressive therapy was reduced; then the patient started to apply topical cidofovir 1-3% twice a day (currently ongoing).

Key message: We report the first Italian case of TS with a whole characterization of the TSPyV in skin, blood and urine samples.