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NEUROFIBROMATOSIS TYPE I: CASE REPORT

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SUMMARY

We present a case of neurofibromatosis type 1 (NF-1) discussing how cutaneous manifestations of disorders or syndromes can be difficult to identify, especially when they are present since birth, present also in a percentage of normal population, and especially when these are isolated findings, not accompanied by other clinical relevant signs or symptoms. It is well known that NF-1 is a rather heterogeneous condition, with different degrees of expression and including cutaneous manifestations. Although there are some life threatening complications clearly associated, for most patients the primary concern and main clinical impact remains the disfigurement caused by the presence and growth of cutaneous-dermal neurofibromas. These lesions can be underestimated especially in the impact on quality of life and sequelae that can cause if abnormally large and not removed. We present a clinical history spanning several years, before managing to properly diagnose our patient with neurofibromatosis type 1 and treat the relevant skin manifestation causing significant deformity and quality of life consequences.

INTRODUCTION

Cutaneous manifestations of disorders or syndromes can be difficult to identify, especially when these are present since birth, when they are seen in a percentage of normal population, and when they are not accompanied by other clinical signs or symptoms. Often the diagnosis can be delayed until a cutaneous manifestation becomes much bigger or symptomatic, or when it changes color or appearance. This can also be the case for café-au-lait spots.

CASE REPORT

A boy of an apparent age of nine years (the parents were not sure about the exact date of birth) was referred to our hospital for a symptomatic skin lesion in his forehead above the left eyebrow that over the last four to five years changed from a flat café-au-lait spot into small pimples surrounding a dark spot of 3 cm in size. No similar cutaneous lesions were present in the rest of the body. The parents told that the mark increased progressively both spreading and also becoming thicker; in addition another bigger mark appeared, also surrounded by small pimples in the left temporal region, measuring 4 X 5 cm in diameter. The lesion had regular margins and was approximately 5 mm elevated; it showed a darker

brown colour, compared to the surrounding skin. Both lesions became progressively symptomatic, with pain and discomfort, which, in the last year or so, became more severe, to an extent that the child could not even sleep and rest properly because of the continuous discomfort. The medical history we collected confirmed that the boy was born with a mark on left side of his head, just above the eyebrow.

The child was then taken, over the last few years, to local dispensaries where the "skin condition" was repeatedly treated without any benefit. Since no improvement was obtained in the local dispensaries, the parents decided to take the child to the District Hospital. Nevertheless, even there a definitive diagnosis was not made. Meanwhile the spot was growing bigger and bigger, affecting the symmetry of his face and displacing also the left ear; in addition it was often quite painful.

With the help of some donors the parents were able to consult a private hospital, where through a CT scan and a skin biopsy, a diagnosis of neurofibromatosis was made. It was clearly communicated that there is no etiologic treatment for the genetic lesion leading to neurofibromatosis.

Nevertheless, the parents were still seeking a treatment for the disfiguring and often painful skin lesions, expanding and growing on the forehead of their child, but they could not find a hospital willing to

attempt any symptomatic surgery or laser treatment.

Finally the boy was referred to Chaaria Mission Hospital and, together with our consultant plastic surgeon, we decided that the operation was possible. Under general anaesthesia we removed the hypertrophic and hyperpigmented skin covering the big mass in the frontal, temporal and orbital regions. After reaching the sub-cutaneous layer, we found that the mass was measuring 10 x 7 cm, was quite soft and infiltrating the surrounding anatomical structures. We realised that it infiltrated the superficial temporal arteries and veins and originated from the auricular-temporal nerve, which was repeatedly twisted, up to showing segmented and "vermiform" aspect with "rosary beads" nodules, which are typical of plexiform neurofibromatosis. The surgery successfully continued with the isolation of the vascular and nervous structures, all the way through their twisted pathway, while preserving the anatomical integrity. Eventually the tumour was completely excised, with particular efforts to maintain the symmetry of the eyes and auricular pinnas.

The tissue removed was again sent for histopathology assessment and the report indicated: "plexiform neurofibroma, virtually pathognomonic of neurofibromatosis type 1".

The patient was discharged with some minimum swelling. He could open and close his left eye unremarkably, and we had repositioned the left ear into its normal position. The surgical incision was made on the line of hair attachment to the forehead, to minimise any impact of the scar. The boy remains well now after seven months, without any asymmetry or residual swelling.

DISCUSSION

Neurofibromatosis type I (NF-1) is a disorder which is caused by a mutation on a gene on chromosome 17 that is responsible for control of cell division. The mutant gene is transmitted with an autosomal, dominant pattern of inheritance, but up to 50% of NF-1 cases can arise de novo, due to spontaneous mutation. The incidence of NF-1 is 1 per 3500 live births.

NF-1 can cause tumours along the nervous system. Common accompanying symptoms of NF-1 include scoliosis, learning disabilities, vision disorders and epilepsy, which in our boy were not present or apparent. Skin conditions, which led to the diagnosis in our patient, are quite frequent in

NF-1 and range from flat, pigmented skin lesions (café-au-lait spots), freckling of the axillae or inguinal region up to dermal neurofibroma. Café-au-lait spots can grow from birth up to 16 years of age; thereafter they are generally not progressive any more. Dermal neurofibroma is manifested as single or multiple, firm, rubbery bumps of varying sizes, with onset generally at puberty. They are progressive in size but not malignant.

CONCLUSION

Surely our young patient will be regularly followed up for the possibility of malignant degeneration and for any possible future skin neurofibroma elsewhere. However the surgery we performed will certainly determine a major impact on his quality of life, with improvement of the stigma the disease had caused.

It is well established that NF-1 is a progressive and heterogeneous condition, with different degrees of expression, making a prognosis difficult to predict.

However, for several NF-1 patients the primary concern remains the disfigurement caused by cutaneous-dermal neurofibromas. Although there is a number of more severe possible complications, most of them are quite rare and many patients with NF-1 live a perfectly normal and unremarkable life.

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