

VON RECKLINGHAUSEN'S DISEASE IN CHILD - A CASE REPORT

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Abstract

Von Recklinghausen Neurofibromatosis is a neuroectodermal dysplasia included in the larger group of phakomatosis; it is an autosomal dominantly transmitted disease with variable penetrance and expression. The authors present the case of a 10 years old female patient with von Recklinghausen disease whose mother has the same pathologic condition. The study estimates somatic development and characteristic clinic manifestations.

Key words: Von Recklinghausen neurofibromatosis, evolution, child.

Introduction

The Recklinghausen disease is included in the family of congenital dysplasia, being characterized by pigmented cutaneous spots (café-au-lait) and benign tumors of the skin, the subcutaneous tissues or perineural (neurofibromes). A positive diagnosis is sustained if more than 6 café-au-lait spots of more than 1.5cm diameter are present. The evolution of the disease is slow; many times patients live a normal life, but often periodical aggravation alternates with stationery ones. Being a hereditary inherited disorder there is no specific treatment for the disease. Continuous follow-up of the patients is important, as possible neurologic complications may appear.

Case presentation

The authors present the patient P.L., female, 10 years old (OD 30791/2004) diagnosed with Neurofibromatosis 1 from the age of 2.

Family history – she has a healthy brother. The mother has the same disease with predominant cutaneous manifestation that appeared gradually after adolescence.

Personal history: the patient is the first born child, from a normal pregnancy, normally delivered at full term; weight at birth – 2400g, height at birth – 48cm, with no neonatal suffering; artificial alimentation from birth; current adequate vaccinations and vitaminizations.

Personal pathologic antecedents: at the age of 8 the patient underwent surgery for excision of a left compressive cervicomedialastinal neuroma. For several months she accuses permanent cephalaea.

At the moment she presents for clinic and paraclinic re-evaluation.

Physical examination findings:

- severe staturo-ponderal deficit – weight=26kg(below 10th percentile) and height=100cm (below 3rd percentile, -4SD);

- multiple flat, café au lait, round or oval spots, disseminated on the trunk and limbs, having various dimensions, between 0.5 and 10cm diameter;
- left laterocervical cheloid scar after surgical excision of the cervicomedialastinal neuroma;
- two soft painless tumors of 0.5 and 1 cm from forehead and the dorsal surface of left foot;
- left palpebral ptosis;
- dorsal cifosis;
- normal mental development.

Paraclinic investigations:

Biological tests: haemoglobin=11,24g%, hematocrit=34%, leukocytes=8500/mm³, segmented neutrophils=63%, eosinophils=4%, lymphocytes=24%, monocytes=6%, thrombocytes=150.000/mm³, ESR (erythrocyte sedimentation rate)=5/9mm, ALT=36U/l, AST=40U/l, urea=23mg%, glycemia=69mg%, seric proteins=6,2mg%, seric calcium= 2,1mmol/l.

Thoracic radiography showed bilateral interstitial reticular and peribronchovascular opacities.

Ventilatory tests- normal values.

Cervical radiography – abnormalities of the C5 vertebra and opacities of the soft tissues at this level.

EEG – rare irritative abnormalities.

Cranial CT-scan found no focal lesions of cerebral substance; the ventricular system was normally positioned, without bone abnormalities.

Slip-lamp examination found iris hyperpigmentation and prominent areas with discoloured border, more numerous in the left eye (Lisch nodules).

Abdominal ultrasonography – normal.

Discussions

Von Recklinghausen Neurofibromatosis is the most common form of the phakomatosis, these being characterised by congenital abnormalities of structures of ectodermal origin like the skin, the nervous system, the retina, and others.

It is inherited as an autosomal dominant trait, but with a highly variable clinical expression among affected individuals within the same family and from one family to another.

A patient meeting two or more of the following criteria can be diagnosed as suffering from Recklinghausen disease:

- A. cafe-Au-Lait Spots (6 or more in 100% of cases)
- pre-Puberty: >5 mm greatest diameter
 - post-Puberty: >15 mm greatest diameter

- B. axillary or inguinal Freckles (2-3 mm diameter);
- C. iris Lisch Nodules (2 or more) ;
- D. optic Glioma (Optic Nerve benign tumor);
- E. neurofibromas of any type (2 or more) or one plexiform neurofibroma;
- F. bone abnormalities ;
- G. first degree relative with positive diagnosis.

Recommendations are to avoid biopsies.

In our case the positive diagnosis was made based on evident clinic manifestations, including pigmented spots, cutaneous fibromas – which meanwhile increased in number and dimensions; of a great importance was the family positive history – mother presenting a cutaneous form of the disease with gradual evolution without any other suffering. Our patient presents a visceral form, having

the compressive mediastinal tumor removed, and also severe development disorders, cervical spine abnormalities, dorsal cifosis, Lisch nodules.

The persistent cephalaea led to cranial CT-scan, but no lesions were found.

EEG shows irritative elements but no seizures were present in history, and also the mental development is adequate.

Conclusions

The two related cases of Recklinghausen disease, mother and daughter, confirm the dominant autosomal inheritance, but with onset of the disease at different ages.

Being a genetic disorder a specific treatment is not available, but the follow-up of the patient is important, as neurological complications are possible.

References

1. Georgescu A, Iagaru N – Modalitati de debut si evolutie in neurofibromatoza la copii. *Pediatria*, 1994, 469-474;

2. Roach SE - Neurocutaneous syndromes. *Pediatr. Clin. North. Am.* 1992, 39-4

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