Paladi Elena, Mariana Sprincean

THE CLINICAL POLYMORPHISM MANIFESTATION IN TUBEROUS SCLEROSIS

IMSP Institutul Mamei şi Copilului (Director – dr. şt. med., conf. univ. S. Gladun)

Key words: Tuberous sclerosis, genetic disorders, hamartomas, developmental delay

Background: Tuberous sclerosis (TS) is a rare genetic disorders with an autosomal dominant pattern of inheritance, variable expressivity that causes benign tumors to grow in brain and other vital organs such as the kidneys, heart, eyes, lungs and skin. It is caused by a mutations of genes TSC1 (encodes for the protein hamartin, located on chromosome 9q34) or TSC2 (TSC2 encodes for the protein tuberin, located on chromosome 16q13.3).

Aim of the study: The importance of clinical and genetic approaches is determined by understanding the disease mechanism for this genetic conditions and it's importace into establish an early clinical diagnostic.

Abstract: A 10 year old girl reported to Central of Reproductive Health and Medical Genetics with seizures. Mother gave a history of mental retardations and epilepsy since 3 year. The patient is under regular treatment for the epilepsy. The girl was born at 39 weeks of gestational age, weighed 3200 g had 52 cm long, a cranial circumference of 32 cm. The clinical examinations revealed white spots on the arms, lumbar region and on the upper part oh the right thorax. Multiple dark macular (confetti-like) lesions and a giant hamartoma formation were seen on the lumbosacral region of the spain.

In family history was found that first child shows similar manifestations but into a milder form.

A provisional diagnosis of Tuberous Sclerosis was established by clinical manifestations. Cerebral magnetic resonance showed hyperostosis of the skull bones and cyst of septum pellucidum. A lumbosacral magnetic resonance showed massive volume formation in soft tissues with lipid-rich content.

Conclusion: It is not uncommon for patients with TS to have symtoms or signs that do not lead to immediate diagnosis. In some cases, the diagnosis is delayed for prolonged period of time. Early diagnosis is very important for through clinical and radiological evaluation, continuous monitoring of symptoms, family planning, genetic councelling and reduction in morbidity and mortality rate.

Dr. Popovici Anca Raluca, Dr. Cobuz Maricela

INFECTIE NOSOCOMIALA LA NOU NASCUT PREMATUR

Se prezintă cazul unui nou-născut prematur, extras prin operație cezariană, vârsta de gestație 29 de săptămâni, care prezintă de la naştere sindrom de detresă respiratorie formă severă, pentru care se instituie suport ventilator şi se administreaza profilactic Curosurf, cu evoluție favorabilă. Ca o complicație a spitalizării prelungite, se constată apariția unei infecții nosocomiale cu germeni de spital (*Pseudomonas aeruginosa* si Streptococ betahemolitic de grup B), care evoluează spre septicemie. Se inițiază tratament cu antibiotic conform antibiogramei, fără complianță terapeutică, motiv pentru care se consideră alternativa continuării tratamentului cu Meronem si Amikacina, ce determină ameliorarea treptată a stării generale şi normalizarea parametrilor de laborator.

Dr. Popovici Anca Raluca, Dr. Cobuz Maricela

HOSPITAL-AQUIRED INFECTION IN A PREMATURE NEWBORN

We present the case of a VLBW premature newborn, first twin, Caesarian section, with a gestational age of 29 weeks that develops since birth severe neonatal respiratory distress syndrome, mechanically ventilated, that received prophylactic Curosurf, with cardio-pulmonary favorable clinical evolution. As a complication, develops hospital-acquired infection with *Pseudomonas aeruginosa* and Group B Streptococcus that escalades to a bloodstream infection. We initiated the treatment under antibiotic sensitivity, without any therapeutic compliance. We considered treatment with Meronem and Amikacin, that slowly improves the clinical status and normalizes laboratory parameters.