**Familial Hypercholesterolemia: A Rare Case of Early Diagnosis**

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Familial hypercholesterolemia (FH) is a common life-threatening genetic condition that causes high cholesterol and leads to a much higher-than-normal risk of coronary heart disease (CHD). The heterozygous type is found in about 1 in 1000 people, whereas the homozygous type is rare in 1 in 10 million people.

**Objective** To analyze the clinical case of family hypercholesterolemia in sibs.

**Patients and Methods** In 3 siblings (from triplets) at the age of 7 years randomly detected plasma LDL cholesterol level of 4.0 mmol/L or higher. In a second study after 14 days, hypercholesterolemia persisted (6.11 mmol/L, 5.67 mmol/L and 6.49 mmol/L, respectively). Two siblings (identical) had a high level of high density lipoprotein (HDL). No evidence of secondary hypercholesterolemia (diabetes mellitus, chronic renal insufficiency, hypothyroidism, hypercholesteremia, iatrogenic illness) was found.

A mother (42 years old), sticks to a strict diet with a reduced fat content, but hypercholesterolemia persists, statins therapy is not conducted, recommendations for examining children have not been received.

**Results** Plasma LDL cholesterol level of 4.0 mmol/L or higher in follow-up blood test, provided parents with hypercholesterolemia, confirms the FH in two children from triplets.

At the same time, there are no external physical signs of the disease in children (xanthomas, corneal arc, xanthelasma). A strict diet with a reduced fat content was recommended to patients, as well as supervision of a cardiologist and lipid screening. It was decided that at the age of 8-10 years, while maintaining LDL cholesterol>4.0 mmol/L in follow-up blood test would be observed even on the recommended diet, the treatment with low doses of statins would be discussed.

**Conclusion** Despite the prevalence of FH and the availability of effective treatment, FH is rarely diagnosed in children. This emphasizes the importance of lipid screening in childhood and cascading screening of all members of the patient’s family for the prevention of CHD.
second group possessed higher values of all parameters in comparison to those from the first and the third groups.

Lesser development of bone tissue revealed by a decreased arm length and size of the major joints probably reflect the anti-androgenic effect of OCPs before and during puberty in males of the first group, while the opposite trend revealed in females from the second group may result from estrogenic effect of mild doses of OCPs. Differences in body mass, waist perimeter and skinfold thickness possessed by volunteers from regions with different environmental conditions may reflect the previously described influence of endocrine disruptor chemicals, such as OCPs, on the balance of leptin, whose secretion is increased in females from the second group may result from estrogenic effect of mild doses of OCPs. Differences in body mass, waist perimeter and skinfold thickness possessed by volunteers from regions with different environmental conditions may reflect the previously described influence of endocrine disruptor chemicals, such as OCPs, on the balance of leptin, whose secretion is increased in females from the second group may result from estrogenic effect of mild doses of OCPs.

The primary goal of this research was to get some insight into knowledge, beliefs and practices of Croatian parents of febrile children. Are they prone to spontaneous reactions caused by an exaggerated fear of fever? To investigate the extent of their ‘fever phobia’, there was a need to ascertain the duration and the height of child’s fever at which parents decided to consult their paediatrician, as well as what possible complications parents associated with high fever. The aim of this study was also to compare beliefs, practices and the sources of information concerning fever management according to the level of the parents’ education and the number of their children.

Parents of 64 febrile children visiting primary paediatrician were interviewed using an anonymous questionnaire with fever-related questions, comprising fever management.

Most parents, 70% of them, claim that high fever is not harmful if treated properly. Children were taken to a paediatrician with fever of 39.1°C by 45% of parents. Although the aforementioned temperature is deemed dangerous by as much as 67% of parents questioned, the less educated parents were more prone to visit the paediatrician even at lower temperatures. Even more worryingly, 33% of all questioned parents came to the paediatrician in under 24 hours of fever’s onset. It should be noted that some of the more educated parents listed more severe possible complications such as brain damage and death of the child; while on the other hand, lowering of the child’s immunity worried more the less educated parents. Even the sources of information on fever management, apart from paediatricians, varied among parents, with educated parents additionally consulting internet sources in contrast to less educated parents who preferred the pharmacist’s advice. No significant difference in parental fever management was observed according to the number of children in the household.

It seems that the proper treatment of fever with children is still insufficiently known to most Croatian parents. The ‘fever phobia’ could cause dilemmas regardless of the level of education of parents who then turn to their health providers for additional advice. The later might deepen the confusion, due to the lack of uniform guidelines and practice for antipyretic treatment of children, especially preschool ones. Further research in this field could lead to better understanding of the problem and possible creation of Croatia’s paediatric guidelines for antipyretic therapy of the febrile child.

**28** FEVER PHOBIA IN CROATIAN FEVER PHOBIA IN CROATIAN PARENTS – WHAT DO THEY KNOW, BELIEVE AND DO WHEN DEALING WITH A FEBTILE CHILD?

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The research in this field could lead to better understanding of the problem and possible creation of Croatia’s paediatric guidelines for antipyretic therapy of the febrile child. Further research in this field could lead to better understanding of the problem and possible creation of Croatia’s paediatric guidelines for antipyretic therapy of the febrile child.

**29** WHEN ONLY BIOPSY CAN PROVIDE AN ANSWER

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**Introduction** Peripheral lymphadenopathy in children is generally benign and self-limited. However, it can be a manifestation of serious underlying disease, so differential diagnosis is essential.

**Case Report** We report a case of a 4-year-old male, previously healthy, presented to the paediatric emergency department with a painless right cervical tumefaction, with about one month of evolution. Fever and weight loss were denied, as well as previous illness and contact with cats. Two antibiotic therapy cycles were performed previously, due to likely bacterial infection, without showing clinical improvement.

On physical examination: Good overall appearance. Rosy face and hydrated.

Right cervical tumefaction (5*3cm, painless to palpation, without local inflammatory signs). Without other palpable lymphadenopathies or hepatosplenomegaly.

Laboratory tests revealed: hemoglobin 11,8g/dL; neutrophil series count 8,74x10⁹/L; platelet count 376 x10⁹/L; no changes in kidney and liver function; C-reactive protein and Erythrocyte sedimentation rate were negatives.

Due to the persistence of tumefaction, hospitalization was decided for etiological investigation. Serological tests for HIV, CMV, Toxoplasmosis and Bartonella henselae were all negative. Tuberculin skin testing was negative.

Chest radiograph showed no changes. Abdominal ultrasonography with two lymphadenopathies of 11 and 15mm in retroperitoneal space, without hepatoesplenomegaly. Cervical ultrasonography revealed a hypocogennic nodular image of regular and well-defined contour in the right jugulo-diagastic chain, measuring 41mmX16mm, and showing no adipose hilum, suggesting lymphadenopathy. Additionally, there were other smaller lymphadenopathies.

Although studies have suggested a benign etiology, due to persistence of tumefaction, a biopsy of the lesion was performed and indicated Burkitt Lymphoma.

**Conclusion** With this work, the authors intend to highlight that if after four weeks of observation and/or empiric therapy, the diagnosis remains uncertain and the lymph node has not regressed in size, a biopsy should be warranted, as only this can confirm or exclude for sure a diagnosis.

**30** A CLINICAL CASE OF A NO EVIDENCE-BASED MEDICAL TREATMENT OF CYTOMEGALOVIRUS INFECTION IN AN INFANT


The diagnosis was confirmed and indicated Burkit Lymphoma.

**Conclusion** With this work, the authors intend to highlight that if after four weeks of observation and/or empiric therapy, the diagnosis remains uncertain and the lymph node has not regressed in size, a biopsy should be warranted, as only this can confirm or exclude for sure a diagnosis.