patient’s survival. This treatment is heavy and expensive, and often it is necessary to repeat it. In Latvia this method of treatment is not available because doctors are not experienced in a wide range of CRS and HIPEC of these rare tumors. There are several clinics in Europe offering this treatment. After treatment, it is important to take control MRI once a year.

**PARENTAL ANALYSIS OF JAG1 GENE IN ALAGILLE SYNDROME**

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**Key words.** Alagille syndrome, genetics, congenital cholestasis

**Introduction.** Alagille syndrome (AS) or arteriohepatic dysplasia is an autosomal dominant (AD) hereditary disorder caused by defects in the components of the Notch signaling pathway, most commonly due to mutations in the JAG1 gene (~97%) or the NOTCH2 gene (~1%). It is a multisystem disorder that primarily affects intrahepatic bile ducts, the pulmonary artery, the heart, the retina, kidneys and has characteristic facial features, but has marked intrafamilial clinical variability. AS has variable expressivity and reduced penetrance (60-96% in various publications). 30-50% of AS patients have an affected parent with mild clinical symptoms, who is referred to as healthy.

**Aim.** The aim of the study was to analyze JAG1 gene mutations in apparently un-affected parents to exclude case of the reduced penetrance, thus, ensuring correct genetic counseling for the family.

**Case description.** A one month old boy with persisting jaundice, pulmonary stenosis, renal dysplasia and retinal hypopigmentation was consulted at Children's Clinical University Hospital. Sequencing of the JAG1 gene was requested due to clinical signs of AS. The results showed an unpublished potentially deleterious sequence variant - an in-frame deletion c.53_73del (p.Leu18_Leu24del) in a heterozygous state.

*Sanger* sequencing of exon I and flanking regions in the JAG1 gene was performed for both parents, but the in-frame deletion c.53_73del (p.Leu18_Leu24del) presented in proband was not found. The mutation was
attributed as *de novo*, providing information for the genetic counseling with slight theoretical probability of gonadal mosaicism.

**Conclusions.** Autosomal dominant diseases with reduced penetrance have to be evaluated and consulted with caution, due to the possible mutation carriers that are clinically unaffected and whose offspring will have a 50% chance of inheriting the mutation. Here we present a case report, where we addressed this problem and obtained information about *de novo* mutation case.

**Summary.** Alagille syndrome (AS) is an autosomal dominant hereditary multisystem disorder due to defects in the Notch signaling pathway. AS has variable expressivity and reduced penetrance (60-96% in various publications). A one month old boy was referred to genetic counseling because of suspected AS due to characteristic clinical signs - jaundice (bile duct paucity), pulmonary stenosis, renal dysplasia, retinal hypopigmentation. Genetic sequencing showed an in-frame deletion c.53_73del (p.Leu18_Leu24del) in the *JAG1* gene in a heterozygous state. *Sanger* sequencing of exon I and flanking regions in the *JAG1* gene was performed for both parents, but the in-frame deletion presented in proband was not found. The mutation was attributed as *de novo*, providing information for the genetic counseling with slight theoretical probability of gonadal mosaicism. It is important to carefully evaluate AD diseases with reduced penetrance and consult patients with caution due to possible clinically unaffected mutation carriers whose offspring will have a 50% chance of mutation inheritance.

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**A PATIENT WITH A FOREIGN BODY IN THE VAGINA AND COMPLICATED POSTOPERATIVE PERIOD**

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**Key words.** Foreign body, Gynaecology, Vesicovaginal fistula

**Introduction.** 17.6% of children presenting with vaginal discharge, have a history of a foreign body in the vagina. Diagnosing a vaginal foreign body in children is challenging because the symptoms are nonspecific and the visual inspection requires general anaesthesia. After the evacuation of the foreign body, up to 30% of patients develop a complication – vesicovaginal fistula (VVF). The aim of this report is to present a girl with vaginal foreign body and