"Edwards Valeo Lifestent avoids redo-surgery of RPA stenosis after repair of Berry's syndrome"

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A 31 • Hypogammaglobulinemia, juvenile myelomonocytic leukemia and macrocephaly : does it fit ?

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INTRODUCTION Juvenile myelomonocytic leukemia (JMML) is characterized by clinical and laboratory features including : splenomegaly, thrombocytopenia, presence of myeloid precursors in peripheral blood, less than 20% blasts in bone marrow, elevated HbF and in vitro hypersensitivity to GM-CSF stimulation. Pathophysiology lies on excessive growth of myeloid progenitors, usually associated with overactivation of Ras pathway. It has been clinically linked to neurofibromatosis type I and Noonan syndrome.

RESULTS We report a 5 months old boy, who presented with enlarged spleen, thrombocytopenia, elevated HbF, presence of myeloid precursors in the peripheral blood. Bone marrow analysis excluded other forms of myeloproliferation including AML, t9,22. Addiional features were macrocephaly with external hydrocephaly, short stature (lower than P3) and low IgG level (0,6 g/L). Though hypersensitivity to GM-CSF was proven, none of the JMML causing genes was mutated. JMML diagnosis was confimed. Later on, he developed refractory hypocalcemia (lower than 7 mg/dl) despite normal vitamin D status. Bone Xray finally allow us to diagnose autosomic recessive malignant osteopetrosis, that recapitulates all clinical signs. Allogenic hematopoietic stem cell transplantation was performed.

CONCLUSION Osteopetrosis (OP) can present as JMML, mainly due to extramedullary hematopoiesis. HbF is also increased in that situation. It is not clear if GM-CSF hypersensitivity is present in majority of OP. As other suggested it, hypogammaglobulinemia is not a feature of JMML and should prompt to reconsider diagnosis. In addition to medullary insufficiency, disturbance of immunoglobulin synthesis worsens immune deficiency in OP. JMML workup should also take into account possibility of osteopetrosis.

A 32 • Verplichting van de opsporing van aangeboren metabole en endocriene ziekten

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INTRODUCTION De neonatale screening naar aangeboren stofwisselingsziekten blijft in Vlaanderen beperkt tot een aanbod, waartegen de ouders zich kunnen verzetten. De auteurs vinden de actuele situatie te vrijblijvend en pleiten voor een algemene verplichting. Het is immers voldoende aangetoond dat de diagnosestelling van deze aandoeningen louter op basis van klinische tekens te laattijdig gesteld wordt, wanneer er reeds irreversiebele letsels opgetreden zijn.

METHOD Onderzoek van mogelijkheid tot algemene verplichting Gezien de nefaste gevolgen van uitstel van de diagnose voor de prognose van de aangetaste neonaat, kan men stellen dat in zijn actuele vrijblijvende vorm aan sommige patiëntjes het recht op een normaal leven ontnomen wordt. Alleen een algemene verplichting, waartegen de ouders zich niet kunnen verzetten op basis van hun ouderlijk gezag, geeft aan alle pasgeborene dezelfde kansen van vroegtijdige opsporing. Anderzijds kan een neonatale opsporing enkel verplicht worden als aangetoond kan worden dat deze inmenging in het privé- of gezinsleven niet als onrechtmatig of arbitrair kan bestempeld worden. Dit kan bezwaarlijk gesteld worden als van het recht op fysieke integriteit van de pasgeborene op het spel staat. In dezelfde zin is het recht op fysieke integriteit de motivatie om aan te tonen in artikel 8 van het Europees Verdrag van de Rechten van de Mens dat deze inmenging een noodzakelijke maatregel ter bescherming van de gezondheid vormt. Hierdoor is het openbaar gezag gemachtigd om zich in het privé- en gezinsleven te mengen. De Belgische Vereniging voor Kindergeneeskunde moet ijveren bij de wetgever voor een algemene verplichting en heeft daarbij een sleutelrol te vervullen.

CONCLUSION Rol van de Belgische Vereniging voor Kindergeneeskunde - De bevoegde overheid overtuigen van een algemene verplichting op basis van de bestaande literatuur. - De algemene bevolking en in het bijzonder de ouders van de pasgeborene grondig informeren omtrent de neonatale screening om de bestaande kennisasymmetrie uit te vlakken. - Het streven naar gelijkvormigheid van het aantal screeningstesten over het ganse land. - Het wetenschappelijk onderzoek promoten om de testmethoden te blijven verbeteren. - De opportuniteit aftoetsen om te screenen naar andere congenitale ziekten.

A 33 • Is early onset adiposity negatively associated with treatment success in obese adolescents

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INTRODUCTION Treatment of childhood obesity is not universally succesfull. Identification of factors associated with treatment failure can help to identify patients groups needing more intensive or tailored follow-up.

AIM . As part of a larger prospective study on the identification of predictors of success or failure of residential treatment in childhood obesity, the aim of this study is to evaluate whether developing overweight at an earlier age (so-called early adiposity rebound) is associated with less weight loss during 9 -12 months of residential treatment.

METHOD The study population consists of 62 adolescents (12-18 year), out of 181, who were treated for obesity in the Zeepreventorium De Haan during 2007-2009 by a multi-component residential program. Inclusion criteria (dutch-speaking, informed consent given, school history traceable) were present in 68 children. Weight and height data at start, during and at the end of treatment were obtained from the files in the Zeepreventorium. By contacting the CLB ("centra leerlingenbegeleiding") growth curves were completed in 62 patients, data from 6 patients could not be traced. Overweight at the age of 6 y was defined by a BMI > 2 Z-score above median for age. Spearman correlation for continuous variables and Fisher exact test for categorical variables were used to test the study hypothesis.

RESULTS No correlation was found between being overweight at the age of 6 and the result of residential treatment at 6 months ($p=0.962$, $r=-0.007$) or at 9-12 months ($p=0.575$, $r=-0.73$). Of the 24 children with a BMI z-score < 2 at 6 year, only one didn't have a normal BMI after treatment. Of the 38 children with a BMI z-score > 2, seven didn't achieve a normal BMI. The difference between these groups is not significant ($p=0.136$). Furthermore no correlation was found between the BMI z-score at 6 years and the percentage of weight loss ($p=0.062$, $r=0.238$). Median weight loss during residential treatment was – 46,6 percent (range +4,4 to – 111%; interquartile range -32 to – 59%).

CONCLUSION : Residential treatment of obesity in the Zeepreventorium is successful. In this small cohort, no association was found between BMI at 6 years and the amount of weight loss during residential treatment.

A 34 • Edwards Valeo Lifestent avoids redo-surgery of RPA stenosis after repair of Berry's syndrome

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INTRODUCTION Severe stenosis of pulmonary arteries are frequently diagnosed after surgical repair of complex congenital cardiac malformations in newborns. Such residual lesions often require multiple catheterizations for pulmonary artery rehabilitation through balloon dilations and stenting. Premounted Edwards Valeo Life stents, designed for biliary tract stenosis, have an open-cell design allowing repeated dilations up to 18 mm, through small (6 Fr) sheats, including in neonates. Such recent stents can accomodate secondary dilations to adult size, thereby avoiding surgery, and preserve the growing potential of vessels between dilations.

RESULTS A full term newborn was admitted with a prenatal diagnosis at 30 weeks with interrupted aortic arch (IAA), aortopulmonary window (APW), and an intra-operative diagnosis of abnormal origin of the right pulmonary artery (RPA), consistent with Berry's syndrome. Since the first case described in 1982 by Berry, less than 30 cases were reported. On the 7th day of life (DOL) we performed a one-stage surgical repair consisting in an end-to-end anastomosis of the aortic arch, patch-closure of the APW and reimplantation of the RPA on the main pulmonary artery (MPA) with a tubular pericardial patch. During follow-up, on the 6th postoperative week, a complete occlusion of the RPA demonstrated by a lung scan and by catheterisation was dilated with coronary balloons up to 4 mm. Three weeks later, an echocardiogram showed re-occlusion of the RPA and an angiography demonstrated an extremely tight stenosis of the RPA origin, dilated with 4 mm Maverick and 5 mm Tyshak balloons. A 6x18 mm low-profile pre-mounted Edwards Valeo Lifestent was positioned and dilated up to 6 mm (fig 2), restoring a normal perfusion of the RPA, as also confirmed by a lung scan and color-Doppler showing no significant residual gradient.

CONCLUSION A newborn diagnosed with Berry's syndrome underwent complete surgical repair on DOL 7 and subsequently developed an extremely tight RPA stenosis 6 weeks later. This complication, likely related to the necessary interposition of a tubular pericardial patch between the RPA and MPA, was successfully treated by balloon dilations and stenting with a 6 mm Edwards Valeo Lifestent. This technique allows repeat dilations up to an adult size RPA (18 mm), thereby avoiding a complex redo-surgery in a newborn.