IMPACT OF HETEROZYGOUS FAMILIAL HYPERCHOLESTEROLEMIA ON LEFT VENTRICULAR MORPHOLOGY AND FUNCTIONS IN PEDIATRIC POPULATION: A SPECKLE TRACKING STUDY

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Background. So far there are few data on cardiac morphology and function in familial hypercholesterolemia (FH) children. Bidimensional strain (S) imaging, has been added to our capabilities and has been proposed as strong index of myocardial contractility. FH children represent a unique clinical model to assess the effect of the pure hypercholesterolemia on cardiac morphology and functions, excluding the effect of co-morbidities. Aim. Thus, we sought to define in FH children the preclinical effects of isolated hypercholesterolemia on the cardiovascular system, by examining left ventricular (LV) function using the more sensitive bidimensional-derived S imaging.

Methods. In all we studied 90 children (45 FH children and 45 controls, mean age 11±3 years). Results. FH children showed thicker LV walls and significantly increased LV mass indexed for height (p=0.0008) and for body surface area (p<0.0001). LV ejection fraction was similar between groups. The diastolic evaluation demonstrated a prolonged deceleration time (p<0.0001), a reduced early diastolic mitral annular velocity (p<0.0001), and higher transmitral early diastolic mitral annular velocity ratio (p=0.0003) in FH children. Longitudinal and circumferential myocardial deformations of the LV were significantly reduced in FH children (p<0.0001). Radial deformation was increased in FH children (p=0.039). Conclusions. This study demonstrated that already during childhood hypercholesterolemia is responsible of significant LV morphological and functional differences when compared to healthy children. Of interest, a decreased longitudinal and circumferential deformations are compensated by increased radial strain in FH children with normal LV ejection fraction. Our data could support the need to initiate cholesterol lowering treatment at a young age in children with FH.
EFFECTS OF ARIPIPRAZOLE AND RISPERIDONE ON VENTRICULAR REPOLARIZATION IN CHILDREN AND ADOLESCENTS

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Background. Atypical antipsychotics (AP) are increasingly being used in children and adolescents for the treatment of psychiatric disorders such as psychoses, autism, attention deficit and hyperactivity disorder (ADHD) and aggressive behavior. Atypical AP may cause QT prolongation on the electrocardiogram (ECG), which predisposes patients to an increased risk of developing threatening ventricular arrhythmias. Although this phenomenon has been exhaustively reported in adults, few studies investigated the safety of these drugs in pediatric patients. We performed an open-label, prospective study to assess the arrhythmic risk of aripiprazole and risperidone in pediatrics.

Methods. Subjects consecutively admitted to the Unit of Child Neurology and Psychiatry of the University of Messina, between January 2009 and December 2012, and starting a treatment of aripiprazole or risperidone, were considered for the study. None of the included patients had a history, signs, or symptoms of cardiovascular, pulmonary or endocrine disorders. A total of 60 patients (55M/5F, mean age 10,2 ± 2,6 years, range 4–15 years), receiving a new prescription of aripiprazole or risperidone in monotherapy underwent a standard ECG before and after two months from the beginning of antipsychotic treatment. Twenty-nine patients were treated with aripiprazole (mean dosage 7.4 ± 3.1 mg/day) and 31 with risperidone (mean dosage 1.5 ± 1 mg/day). A standard 12-lead ECG was obtained for each patient with the same instrument (Cardioline delta 3 plus). The duration of QT intervals was measured manually in all leads in which the onset of the QRS complex and the return of T wave to baseline were clearly identified. The QT duration was corrected for heart rate according to the Bazett's formula to produce QTc and averaged for all assessed leads (mean QTc). If respiratory sinus arrhythmia was present the QT interval was measured in all leads where RR intervals were almost equal. Heart rate, RR, PR and QRS intervals were also measured. QTd was calculated as the difference between the longest and shortest individual lead QTc. QTc intervals > 450 msec or 60 ms longer then at baseline, and QTd > 100 ms were considered abnormal. Basal and post-treatment ECG parameters, including QTc and QTd interval duration, were compared within treatment groups. Mann–Whitney U-test was used for unpaired group comparison and Wilcoxon paired rank test was used for comparisons across time. Two sided tests were used and statistical significance was accepted at a p level <0.05.

Results. Although none of the patients exhibited pathological values of QTc or QTd, treatment with risperidone was associated with a significant increase of both QTc and QTd values (407.4 ± 11.9 ms vs 412.4 ± 10.3 ms, p<0.05; and 40.0 ± 4.4 ms vs 44.7 ± 5.5 ms, p<0.01, respectively). Conversely, treatment with aripiprazole was associated with a statistically significant increase of QTd, (40.6 ± 6.5 ms vs 46.3 ± 7.2 ms, p<0.01) in presence of an unmodified QTc. Moreover in patients treated with aripiprazole a significant decrease in heart rate was observed after treatment (90.0 ± 21.2 bpm vs 79.8±19.3 bpm; p<0.01).

Conclusions. This study underlines the relative cardiac safety of aripiprazole and risperidone in childhood, even if these drugs may exert an effect on ventricular repolarization. However, a particular caution in prescribing these drugs should be exercised by physicians in patients with a genetic predisposition to arrhythmias. It might be useful to execute an ECG examination in all patients undergoing AP therapy. Additionally, both QTc and QTd should be measured in order to warrant a reliable assessment of drug-induced QT prolongation.
RHABDOMYOMAS AND TUBEROUS SCLEROSIS: OUR EXPERIENCE IN 27 CASES

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Introduction
Rhabdomyomas are the most frequent cardiac tumors in children and are often associated to tuberous sclerosis complex (TSC) (1). This is an autosomal dominant neurocutaneous disorder that can affect most commonly the brain, kidneys, heart, and lungs (2). We report our experience with regard to 27 patients with multiple cardiac masses.

Patients and Methods
From January 1989 to December 2012 we diagnosed Rhabdomyomas in 27 babies. In six cases (22 %) the masses were detected antenatally, in seven cases (26 %) during first three days of life, in others seven cases (26 %) the patients were two months old and the last seven children (26 %) were subjected to echocardiography before third and twelfth months of life. In 17 patients (62%) we detected tuberous sclerosis confirmed by genetic test for TSC. Postnatally (78 %) the indication for echocardiography was in 2 cases (7,5 %) an arrhythmia, in 12 cases (44,5 %) the presence of heart murmur, in 3 cases (11 %) the appearance of seizures like infantile spasms, whereas in 4 cases (15 %) the echocardiographic examination had been performed as a screening. In 24 (89 %) patients we detected multiple mass that led us to define these masses as rhabdomyomas. In three patients all with TSC (11 %) we observed only a single cardiac mass. Subsequently to the diagnosis, all babies were subjected to EEG and MRI of brain. The family history was investigated. All babies were monitored through the execution of ECG and echocardiography-color Doppler every six months and of Holter-ECG every twelve months.

Results and Considerations
Only in 3 babies (11 %) the mass localized in right or left ventricular outflow tract caused a significant obstruction. One newborn (4 %) with diagnosis of cardiac mass in fetal period, as she showed signs of heart failure and cyanosis, at birth was subjected to surgery to remove the mass. In 1 (4 %) patient echocardiography examination revealed only slight hypokinesia without need medical or surgical therapy. Whereas one (4 %) antenatally diagnosed died soon after birth because of severe hypokinesia. The other patients (77 %) were totally asymptomatic. In 6 cases (22 %) we found atrial and/or ventricular ectopic beats and in 2 (7,5 %) of these cases it was revealed random and constituted an indication to echocardiography. For all patients we have not considered necessary a drug treatment and the arrhythmia is spontaneously totally disappeared or has been gradually reduced. Only in 1 (4 %) Wolf-Parkinson-White syndrome was diagnosed. Familial history of disease resulted positive in six cases (22 %). With regarding to peculiar skin features in neonatal age, 9 (33 %) showed hypomelanotic macules.

All infants with MRI typical TSC 17 (62%) lesions developed seizures during follow-up and we could not rule out also the other patients will develop subsequently.

During follow-up we observed in all 26 survived a reduction of rhabdomyomas in terms of both number and size. Despite the potentially favorable cardiac evolution of patients with cardiac rhabdomyomas, their presence suggest a tuberous sclerosis with a neurological prognosis that is not related to the number or the dimensions of rhabdomyomas. In our series only patients with severe symptoms at diagnosis had a poorer cardiac prognosis.

References
VALVOLA AORTICA BICUSPIDE E PERSISTENZA DELLA VENA CAVA SUPERIORE SINISTRA: UNA DIAGNOSI ELETTROCARDIOGRAFICA?

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PREMESSE E SCOPI
La vena cava superiore sinistra persistente (VCSSP) si può ritrovare in circa lo 0,4% delle persone. Spesso è isolata ma in circa il 10% dei casi può associarsi ad anomalie congenite come canale atrioventricolare, tetralogia di Fallot, DIA, DIV, ritorno venoso anomalo polmonare, coartazione aortica e valvola aortica bicuspide (VAB). Inoltre, la VCSSP può essere associata ad anomalie strutturali del tessuto di eccito-conduzione con relative alterazioni del ritmo tra cui ritmo del seno coronarico, sindrome di Wolff-Parkinson-White e malattia del nodo del seno. I disturbi del ritmo sono legati anche alla dilatazione del seno coronarico, che determina uno stiramento del miocardio atriale e del tessuto di conduzione. La VCSSP è di solito l’unica vena cava superiore (assenza di vena anonima) ma talora è associata a una vena cava superiore destra, con cui comunica tramite una vena anonima ipoplasica. La VCSSP drena quasi sempre nell’atrio destro tramite un seno coronarico (SC), dilatato per il flusso indebito; solo raramente drena nell’atrio sinistro a causa di un SC unroofed. Scopo del lavoro è stato valutare l’associazione tra la VCSSP, cardiopatie congenite e anomalie del ritmo.

MATERIALI E METODI

RISULTATI
Si sono individuati 17 pazienti con VCSSP (10 maschi, 7 femmine), di età compresa tra 1 e 30 anni (età media 7 anni). L’anomalia vascolare era così associata: 1 pz con VAB, coartazione aortica e ritmo del seno coronarico; 2 pz con VAB e ritmo del seno coronarico; 1 pz con VAB e coartazione in sindrome di Turner; 2 pz con VAB e coartazione aortica; 1 pz con VAB, coartazione aortica e anomalia coronarica; 1 pz con VAB, interruzione dell’arco e DIV subaortico; 1 pz con VAB, DIV muscolari e displasia polmonare; 1 pz con valvola di Eustachio gigante; 1 pz con DIV subaortica; 1 pz con DIA ostium II; 1 pz con tetralogia di Fallot; 4 VCSSP isolate.

CONCLUSIONI
In questo studio si riscontra una frequente associazione tra persistenza della vena cava superiore sinistra e VAB (53%); inoltre, il 33 % dei pazienti con bicuspidia aortica presenta un ritmo atriale ectopico (del seno coronarico). Nonostante il bias di selezione questi dati sono interessanti. La valvola aortica bicuspide è un’anomalia non rara (prevalenza 1%), e non sempre minore. Conoscerne l’esistenza è importante, ma esistono forme che possono eludere un primo ecocardiogramma; quindi un seno coronarico dilatato o la visualizzazione diretta di una VCSSP possono richiamare maggiore attenzione sull’anatomia valvolare aortica e sulle altre condizioni potenzialmente associate. Il ritmo atriale basso, o del seno coronarico, viene solitamente considerato benigno e fisiologico. In questa casistica tale ritmo presenta una considerevole associazione con VCSSP e valvola aortica bicuspide. Il dato non appare casuale ed è comprensibile alla luce degli aspetti anatomici sopradetti. Si può dunque affermare che il riscontro di uno o più ritmi atriali bassi debba essere seguito da un ecocardiogramma.
DETECTION OF LEFT VENTRICULAR DYSFUNCTION BY TISSUE DOPPLER IN ASYMPTOMATIC PATIENTS WITH HISTORY OF SURGERY FOR CONGENITAL HEART DISEASE

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Background: Patients with history of surgery for congenital heart disease often show apparently normal left ventricular (LV) function when assessed by standard systolic and diastolic indexes such as ejection fraction and transmitral flow pattern. Assessment of long-axis LV dynamics by tissue Doppler provides early and sensitive indexes of LV performance that can unmask LV systolic and diastolic dysfunction despite normality of standard indexes. However, the existence of subtle abnormalities in LV function in these patients has never been explored.

Methods: Thirty-seven patients with previous surgical correction of congenital heart disease and no clinical evidence of heart failure (status post [s/p] group), and 37 age-, gender- and body surface area-matched healthy controls were recruited. Most common congenital lesions leading to surgery in the s/p group were tetralogy of Fallot (n=13, 35.1%), transposition of the great vessels (n=5, 13.5%), aortic coarctation (n=5, 13.5%), ventricular septum defect (n=4, 10.8%), and atrial septum defect (n=4, 10.8%). All subjects underwent echocardiography to assess standard indexes of LV function. Tissue Doppler was used to assess LV peak longitudinal velocity at systole (s’) and early diastole (e’). Values were calculated by averaging measurements taken at the septal and lateral site of the mitral annulus. The ratio of peak early diastolic LV filling velocity to e’ was also calculated and used as an index of LV filling pressure.

Results: As a result of 1:1 matching procedure, mean age was 8.5 ± 4.8 in both groups, with equal gender distribution (45.9% female) and similar body surface area (1.0 ± 0.4 m² vs 1.1 ± 0.4 m², p=0.74). The s/p group and the controls showed no differences in indexed LV mass (63.2 ± 19.9 g/m² vs 58.4 ± 21.6 g/m², p=0.32), LV ejection fraction (63.0 ± 7.0% vs 63.8 ± 4.5%, p=0.53), mitral E/A ratio (2.2 ± 0.9 vs 2.1 ± 0.6, p=0.46), and E wave deceleration time (143 ± 37 ms vs 146 ± 35 ms, p=0.66). However, s/p patients showed depressed s’ (8.0 ± 1.5 cm/s vs 9.4 ± 1.8 cm/s, p=0.0004) and e’ (13.7 ± 2.1 cm/s vs 15.6 ± 2.0 cm/s, p<0.0001) and higher E/e’ ratio (6.9 ± 2.3 vs 6.1 ± 1.1, p=0.039) in comparison with the controls. Thirty-six (97.3%) patients in the control group, but only 29 (78.4%) in the s/p group, showed an E/e’ ratio in the normal range (<8). Similar findings were obtained when the analysis was focused to the subgroup with history of corrected tetralogy of Fallot.

Conclusions: Despite normality of LV mass, ejection fraction, and transmitral flow pattern, patients with previous surgical correction of congenital heart disease show significant LV systolic and diastolic dysfunction compared to matched controls. This impairment seems to be present even when the underlying congenital disease is expected to predominantly involve right-sided heart chambers. Considering the established prognostic importance of LV long-axis impairment, careful clinical and echocardiographic follow-up, including assessment of longitudinal LV function by tissue Doppler, should be considered mandatory in these patients.
Background: Rheumatic heart disease (RHD) affect 15.6–19.6 million people worldwide and represent a common cause of mortality and morbidity. RHD leads to disability and premature death, with important economic and social consequences. The few studies performed in Africa showed a prevalence of RHD in school children varying between 1.4-6.2 % and demonstrated that the minor rheumatic valve lesions could not be detected by the only clinical criteria.

Patients and methods: From January 2012 to April 2013, four groups of operators, each consisting of a cardiologist and a nurse, performed 5 missions in Eritrea for screening and prevention of RHD. Each group had a portable echocardiography devices, a power generator, a laptop computer, light bulb for throat examination, stethoscope, rapid tests for diagnosis of streptococcal infection in progress, oral antibiotics. According to Eritrean Ministry of Health and Education, we worked together with local doctors and nurses in order to form them.

We randomly selected students from 51 different schools of Asmara (elementary and junior) for a total of 3765 student (M 1872, F 1893) with mean age 10 years (range 5-18 years).

Students received a simple lesson with few slides on prevention, explaining what is RHD and how to prevent it, which are the symptoms of streptococcal tonsillitis and how to treat it. Next step was to collect medical history regard to pharyngeal tonsillitis, tonsillectomy or other major diseases, and finally we made complete medical examination.

Ultrasound heart study was performed in each student. At present no clear echocardiography criteria exists for the early diagnosis of RHD. According to latest recommendations, moderate thickening of valve leaflets was considered an indicator of established rheumatic heart disease.

Results: We found red pharynx in 43 students (1.1%). All of them underwent a rapid test for group A streptococcal research with 7 positive results. Oral antibiotics were administered to children with infection on progress.

In 119 students (3.1%) we found signs of valvular damage as detailed in table 1.

Table 1: Incidence and severity of rheumatic heart disease among Eritrean school children.
AO=aorta; MV=mitral valve; RHD=rheumatic heart disease.
Students with normal heart, have been educated to primary prevention, while students with ultrasound exam suggestive for RHD were referred to the local pediatric cardiologist, to start an adequate follow-up and secondary prophylaxis.

The local staff was been properly instructed to perform the screening autonomously in order to continue the program when we will complete our work.

Of interest, as an additional data, in 41 students (1%) various congenital heart defects were also diagnosed. The most significant ones have been reported to the local pediatric cardiologist.

Conclusion: Data on RHD in Africa are few and the incidence of this disease in some areas like Eritrea is not known. The African population is exposed to the effects of streptococcal pharyngitis for poor hygiene, genetic predisposition and poor prevention. The data we have collected so far are related to the urban area of Asmara, therefore we expect to find a higher incidence of RHD outside the town.

Early detection of subclinical RHD is vital, because prophylactic penicillin can prevent progression to clinical valve disease and heart failure. Secondary prophylaxis, have been shown to be inexpensive and efficient and has been recommended by the World Health Organization and the World Heart Federation since the 1980s. Children education to primary prevention and accurate instruction of local staff are essential for long term success of a prevention program for RHD.
IL FLUSSO IN VENA CAVA SUPERIORE COME MISURA DELLA PERFUSIONE SISTEMICA NEL NEONATO CRITICO: PROPOSTA DI UNA NUOVA TECNICA ECOGRAFICA

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Introduzione
Una promettente metodica di monitoraggio della perfusione sistemica nel neonato prematuro è la misurazione ecografica del flusso in vena cava superiore (VCS), che rappresenta il flusso di sangue refluo dalla parte superiore del corpo, l'80% circa dal cervello. Un ridotto flusso in VCS è associato infatti allo sviluppo di emorragie intraventricolari.

La tecnica tradizionale prevede la misurazione del diametro della vena da una proiezione parasternale asse lungo e la misurazione della velocità del flusso (VTI) da una proiezione sottocostale. Dal diametro si calcola l'area, assumendo la vena perfettamente circolare. Moltiplicando l'area per il VTI e la frequenza cardiaca, si ottiene il flusso.

Tale metodica presenta però una grande variabilità tra osservatori, che ne limita la diffusione nella pratica clinica. Per tale motivo proponiamo una nuova tecnica che prevede di misurare direttamente l'area della VCS da una proiezione trasversale, a livello della biforcazione dell’arteria polmonare (v. Figura1).

In tal modo si evita di introdurre un errore sistematico calcolando l'area dal diametro, data la forma non circolare della vena. Inoltre proponiamo di misurare il VTI da una proiezione sovra-sternale. In tal modo i movimenti respiratori e il meteorismo addominale non ostacolano l'acquisizione delle immagini.

Obiettivi dello studio
Gli obiettivi dello studio sono:
- confrontare in 10 neonati le tecniche ecografiche nuova e tradizionale con la Risonanza Magnetica a contrasto di fase (PCMRI)
- verificare in 20 neonati la variabilità intra-osservatore tra due misurazioni del flusso in VCS secondo le due tecniche ecografiche

Metodi
I pazienti studiati con ecografia e PCMRI sono stati reclutati da giugno a novembre 2012 presso le Patologie Neonatali dell’Hammersmith Hospital, Imperial College e del St. Thomas’ Hospital, Kings College, London. È stato utilizzato un ecografo Vivid 7 (GE Healthcare). L'ecocardiografia è stata eseguita entro ventiquattro ore dalla PCMRI, nelle stesse condizioni di sonno o veglia tranquilla. La PCMRI è stata eseguita con scanner 3T (Phillips), senza sedazione, durante il sonno naturale, previo posizionamento di protezioni acustiche.

I pazienti studiati solo con ecografia sono stati reclutati da dicembre 2012 a marzo 2013 presso la Patologia Neonatale dell’Azienda Ospedaliera Universitaria Integrata di Verona. È stato utilizzato un ecografo Vivid 7 con le stesse modalità di analisi delle immagini.

Risultati e conclusioni
I dieci pazienti sottoposti a ecografia e PCMRI presentavano un’età media di 36+3 settimane e un peso di 2123g.
L'area della VCS calcolata dal diametro, secondo la tecnica tradizionale, sottostima l’area della vena misurata in PCMRI (13 vs 28 mm2) con un bias medio di -15 mm2 e una scarsa correlazione ($r^2 0.101$).
L’area misurata con la tecnica nuova mostra invece una migliore correlazione con l’area in PCMRI ($r^2 0.603$) e un bias medio di solo -3 mm2.
Entrambe le tecniche ecografiche sovrastimano la velocità di flusso in VCS rispetto alla PCMRI, per un limite intrinseco dell’ecografia nel determinare la velocità di un fluido in moto turbolento. La misurazione da una proiezione sovra-sternale offre però una stima più accurata rispetto a quella sottocostale (bias medio +6 vs +11 cm).
Di conseguenza, il flusso in VCS con la tecnica tradizionale mostra una scarsa associazione con la PCMRI ($r^2 0,254$), mentre la tecnica nuova risulta più accurata ($r^2 0,657$).

La coorte di 20 pazienti sottoposta a due misurazioni ecografiche ripetute del flusso in VCS secondo le due tecniche presentava un’età media di 32 settimane e un peso di 1557g. Secondo l’analisi di Bland Altman la tecnica nuova mostra una ridotta variabilità intra-osservatore, con coefficiente di ripetibilità che, da 50 ml/kg/min nella tecnica tradizionale, viene dimezzato a 25ml/kg/min.

In conclusione la nuova tecnica ecografica sembra essere uno strumento più affidabile e accurato per identificare i neonati a rischio di ipoperfusione e di sequele neurologiche severe.
VALUTAZIONE DELLA FUNZIONE VENTRICOLARE NEI NEONATI CON STRAIN E STRAIN RATE:
INDIVIDUAZIONE DEI VALORI DI normalità COME PRIMO PASSO VERSO L’APPLICAZIONE CLINICA

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SCOPO DELLA RICERCA. 2D speckle tracking (2DSTE) strain and strain rate (SR) sono tecniche poco usate nei neonati per quantificare la funzione ventricolare e a tutt’oggi mancano valori normali di riferimento in questa popolazione. Il scopo di questo studio è individuare valori di 2DSTE strain and SR in neonati sani.

METODO IMPIEGATO. Abbiamo arruolato in modo prospettico 68 neonati sani a termine, con età < 72 ore. Strain e SR longitudinali sono stati ottenuti dalle proiezioni 4-,2-,3-camere apicale per il ventricolo di sinistra (VS) e dalla 4-camere apicale per il ventricolo di destra (VD). Strain and SR radiale del VS sono stati ottenuti dalla parasternale asse corto a livello apicale, medio-ventricolare e basale. Sono stati archiviati 3 cicli cardiaci per l’analisi off-line (EchoPAC version 7.0 GE system). Tutte le misurazioni sono state fatte da un operatore diverso da chi ha fatto le acquisizioni. Un secondo osservatore cieco ha analizzato 15 casi per valutare la variabilità interosservatore. I valori sono espressi come media ± DS. Il confronto dei valori di strain and SR nei diversi segmenti del VS e del VD è stato ottenuto con l’analisi di varianza con la correzione di Bonferroni per test multipli (p <0.05)

RISULTATI E CONCLUSIONI. I valori di riferimento di strain and SR sono riportati nelle tabelle 1 e 2. La riproducibilità tra gli osservatori è stata più elevata per lo strain e SR radiale piuttosto che quello longitudinale. Il nostro studio dimostra prospetticamente in una numerosa popolazione di neonati sani che i valori di strain e SR longitudinali non sono omogenei e sono caratterizzati da valori più bassi per il VS che per il VD. Al contrario i valori di strain e SR radiale sono più omogenei e più alti per entrambi i ventricoli rispetto ai valori longitudinali. Questo studio ha individuato valori di riferimento per neonati, utilizzabili per ulteriori studi di funzione cardiaca in questa particolare popolazione usando le nuove tecniche di 2DSTE strain and SR.
CLINICAL OUTCOME, VALVE DYSFUNCTION AND PROGRESSIVE AORTIC ENLARGEMENT IN A PEDIATRIC POPULATION WITH ISOLATED BICUSPID AORTIC VALVE

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Aim of study: Bicuspid aortic valve (BAV) is the most common valvular congenital anomaly, affecting 0.5-2% of the general population. Although it’s generally considered a benign condition, concern has been raised in the last decades about its association with progression of valve dysfunction and progressive aortic enlargement. Since the large majority of studies concerning natural history and complications related to BAV have been conducted in adults, the clinical outcome and the risk of progression of valve dysfunction and aortic dilation in pediatric patients (pts) have still to be defined. The aim of this study was to explore the long-term clinical outcome and the risk of progression of aortic valve disease and aortic dilation in a pediatric population with isolated BAV observed in Cardiology Unit at Pediatric Hospital A. Meyer in Florence.

Methods: A total of 179 pediatric pts (median [95% IQ] age at enrolment 7.8 [2.7-12.0] years, 76.5% male) with isolated BAV were prospectively followed. Pts with severe valve stenosis or regurgitation at baseline were excluded. The clinical endpoint included cardiac death, aortic complications and need for surgery or percutaneous valvuloplasty. The echocardiographic endpoints were: 1) progression of aortic stenosis (AS) or regurgitation (AR) from baseline to follow-up, defined as an increase by 1 grade according to a three-level scale severity; 2) significant progressive aortic enlargement at different levels of the aortic root, defined as development of a Z-score >2 not present at baseline.

Results and conclusions: At the initial observation, AS and AR were present in 25 (14.0%) and 87 (48.6%) pts, respectively. After a median follow-up of 5.4 [2.3-9.2] years, all pts were alive. The clinical endpoint occurred in 4 (2.2%) pts (0.41 events/100 patient-years): elective valvuloplasty was performed in 3 pts for severe AS and 1 patient underwent aortic valve replacement for severe valve dysfunction (AS and AR). AS was present in 28 (15.6%) pts, whereas AR was present in 102 (57.0%). A progression in AS and AR was observed in 9 (5.0%) and 29 (16.2%) pts, respectively. The Z-scores at the end of follow-up were not significantly different from those observed at baseline at the annulus, Valsalva sinuses and sinotubular junction, whereas a slight increase was observed at the level of the ascending aorta (1.9 vs 1.5, p=0.046). Significant progressive aortic enlargement occurred in a minority of pts (10.6%, 5.6%, 9.5%, and 19.0% respectively). No baseline variable was able to significantly predict the clinical endpoint, the progression of AR, or progressive aortic enlargement at the level of annulus, sinotubular junction, or ascending aorta. A low Z-score at the aortic annulus was the only variable predicting progression of AS (odds ratio 0.59, p=0.014), whereas fusion of the right and left coronary cusps was the only predictor of progressive enlargement of Valsalva sinuses (odds ratio 7.18, p=0.046). In conclusion, the clinical outcome in paediatric pts with isolated BAV is favourable and the progression of aortic valve dysfunction and aortic enlargement is relatively slow during childhood and adolescence. On the other hand, considering that a significant progressive enlargement occurred in the ascending aorta, albeit in a minority of pts, our data might justify the need for a careful echocardiographic follow-up in all these pts, even in those with normal aortic diameters at first diagnosis.
TOTAL ANOMALOUS PULMONARY VENOUS CONNECTION: IS ECHOCARDIOGRAPHIC DIAGNOSIS SUFFICIENT IN ALL CASES? OUR 24 MONTHS EXPERIENCE

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BACKGROUND: Total anomalous pulmonary venous connection (TAPVC) can be isolated or complex type -when associated to other intra-cardiac lesions besides atrial septal defect and patent ductus arteriosus-. It is frequently misdiagnosed and without operative correction early death is common. It is important to achieve a correct and precise preoperative diagnosis which is usually accomplished by echocardiogram and sometimes by angiography, this last even if invasive, been considered the gold standard until the advent of cardiac magnetic resonance angiography (MRA) and computed tomography angiography (CTA).

AIM: Aim of our study was to review our 24 months experience with diagnosis of TAPVC both isolated and complex type and to demonstrate that even in complex cases echocardiographic data are sufficient to achieve a correct diagnosis of the cardiac disease also in complex and mixed type of TAPVC. In a few, more complex cases, CTA can be very helpful to directly demonstrate some connections in order to precisely plan surgical correction.

METHODS AND MATERIALS: We retrospectively reviewed TAPVC cases treated in our center in 24 months, from April 2011 to April 2013. We collected data from 10 patients. Mean age at diagnosis was 7 days (1-35 days), mean weight was 2 Kg (0.8-3 kg). CTA was performed in 5 patients. Cardiac catheterization was done in 0 cases. Surgical correction was achieved in all cases.

RESULTS: TAPVC was isolated in 3 cases, part of asplenia syndrome in 4 cases, associated to other cardiac lesions in 2 cases (ventricular septal defect, aortic arch hypoplasia) and 1 associated to tracheo-oesophageal fistula. TAPVC was mixed type in 2 case, totally infracardiac in 7 cases and totally supracardiac in 1 case. Echocardiographic diagnosis were confirmed in 5 patients by surgery, in 3 patients by CTA and later on by surgery. In two cases of mixed type CTA added some information which were useful for surgery. Echocardiographic and CTA diagnosis –when obtained- were confirmed in 100% of cases by surgical inspection. Mortality at surgical time was 0%. Mortality at one month follow up was 10% (1 asplenic syndrome with mixed type TAPVC). No added mortality were recorded ad 6 months follow up where done.

CONCLUSION: Though CTA can facilitate or better demonstrate TAPVC diagnosis especially in more complex mixed type cases, echocardiography alone is in most cases sufficient to accomplish a precise diagnosis even in complex subgroups and can correctly guide surgical correction and follow up in this congenital heart disease. Cardiac catheterization in this setting of patients is no more indicated.
STUDIO ECOCARDIOGRAFICO DELLA VALVOLA AORTICA BICUSPIDE IN UNA POPOLAZIONE PEDIATRICA

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BACKGROUND: la valvola aortica bicuspide (BAV) rappresenta la più comune cardiopatia congenita con prevalenza pari al 0.5-2%; colpisce il sesso maschile più del femminile con rapporto 3:1; è associata a rischio significativo di sviluppare aneurisma dell'aorta toracica e dissezione aortica.

SCOPO DEL LAVORO: studiare la relazione tra morfologia e funzione della BAV in una popolazione pediatrica non selezionata.

MATERIALI E METODI: da settembre 2010 ad aprile 2013, sono stati sottoposti ad ecocardiografia 1716 soggetti di età compresa tra 4 e 18 anni (mediana 12 anni) riferiti all'ambulatorio di ecocardiografia da pediatri di libera scelta (52%), medici dello sport (37%) e cardiologi dello sport (11%) per approfondimento diagnostico dopo visita di routine o visita medico-sportiva per idoneità agonistica. Tutti gli esami sono stati effettuati da due operatori, un cardiologo ed un medico dello sport, con ecografo Siemens Acuson CV70 munito di sonda pediatrica p9-4 e da adulto p2-4. Per lo studio della valvola aortica sono state utilizzate proiezioni parasternale asse lungo e asse corto, particolarmente favorevoli per la ricerca e quantificazione dei lembi valvolari. Secondo le Linee Guida ASE 2005 le dimensioni aortiche sono state misurate a livello dell'Anulus, dei Seni di Valsalva, della Giunzione Sino-Tubulare e dell'Aorta Ascendente. Per lo studio della funzione aortica, i difetti valvolari (stenosi ed insufficienza) sono stati quantificati in accordo con le Linee Guida SIEC 2011.

RISULTATI: La BAV è stata diagnosticata in 39 pazienti (22%), con rapporto M:F pari a 2,7:1 (28:11). La rima di chiusura valvolare è risultata orizzontale (BAV tipo A) in 28 casi (71%) e verticale (BAV tipo B) in 11 casi (29%). Nel 69% dei casi il rafe si trovava tra la cuspidi coronarica destra e la coronarica sinistra. Il 62% di pazienti (n. 24) presentava insufficienza valvolare isolata e di grado lieve; di questi il 67% (n.16) presentava BAV tipo A. Nel 5% dei soggetti (n.2) è stata riscontrata dilatazione dell'aorta ascendente associata ad insufficienza valvolare di grado moderato; anche in questo caso la BAV era di tipo A. Il 13% dei pazienti (n. 5) presentava steno-insufficienza valvolare e di questi il 60% (n .3) era affetto da BAV tipo B con rafe tra la cuspidi coronarica destra e la non coronarica. Il 20% (n.8) dei pazienti non presentava disfunzione valvolare aortica. Particolare attenzione è stata dedicata a 2 fratelli, riferiti all'ambulatorio per screening ecocardiografico in considerazione della forte familiarità per BAV (sia madre che padre): solo il minore, maschio di anni 5 è risultato positivo per BAV tipo B, il maggiore, maschio di anni 9 è risultato negativo.

CONCLUSIONI: nella valutazione ecocardiografica di una popolazione pediatrica merita particolare attenzione lo studio morfologico e funzionale della valvola aortica. Il nostro studio afferma che la più frequente disfunzione valvolare associata a BAV tipo A è l'insufficienza isolata, mentre il tipo B è con maggior prevalenza associato a steno-insufficienza aortica.
PERCUTANEOUS TREATMENT OF AORTIC COARCTATION: THE VALUE OF PHYSICAL ECHO STRESS IN THE FOLLOW UP

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Background: An important functional parameter in the assessment of the effects of percutaneous repair of CoA is the maximal pressure gradient in the descending aorta with the presence of diastolic run off. Increasing of these parameters after exercise stress test is a clinical parameter that need to be evaluate. Aim of the study was to evaluate the echocardiographic maximum gradient after physical stress as indicator of recoarctation in patients underwent to stent implantation.

Methods: In the last 2 years 30 patients underwent to stent implantation for primary aortic coarctation. All of them were evaluated at 6 months from the percutaneous procedure with ecocardiography at rest and after physical stress. Of these, 10/30 (33%). (8 males, 2 females; mean age 21.8+ 5.4 years) showed a significative increase of the maximum gradient. Therefore in this subgroup a second percutaneous procedure was indicated. Echocardiographic maximum gradient in the descending aorta at rest and after physical stress were evaluated and then compared with catherlab data.

Results: At the rest echo the mean value of maximal pressure gradient was 45mmHg +/- 4 mm Hg without diastolic run off while at the end of the physical stress was 83mmHg +/- 13mmHg (P <0.02 versus rest) with olodiastolic run off. In the cath lab we found a mean peak to peak gradient of 25 mmHg +/- 5mmHg . In 5/10 patients (50%) we performed a stent redilatation.

Conclusions: Physical stress echocardiography could be a predictive exam for recoarctation even if in about 50% of the cases it overexitmate the gradient
DETECTION OF RIGHT VENTRICULAR DYSFUNCTION BY TISSUE DOPPLER IN ASYMPTOMATIC PATIENTS WITH HISTORY OF SURGERY FOR CONGENITAL HEART DISEASE

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Background: Poor data exist about the long-term effect of surgery on right ventricular (RV) dysfunction in asymptomatic patients with congenital heart disease submitted to surgical correction.

Methods: Thirty-seven patients with previous surgical correction of congenital heart disease and no clinical evidence of heart failure (status post [s/p] group), and 37 age-, gender- and body surface area-matched healthy controls were studied by echocardiography. Most common congenital lesions leading to surgery in the s/p group were tetralogy of Fallot (n=13, 35.1%), transposition of the great vessels (n=5, 13.5%), aortic coarctation (n=5, 13.5%), ventricular septum defect (n=4, 10.8%), and atrial septum defect (n=4, 10.8%). M-mode was used to assess tricuspid annulus plane systolic excursion (TAPSE), and pulsed tissue Doppler was used RV peak longitudinal velocity of tricuspid annulus at systole (s') and early diastole (e'). The ratio of peak early diastolic RV filling velocity to e' was also calculated and used as an index of RV filling pressure.

Results: Mean age was 8.5 ± 4.8 in both groups, with equal gender distribution (45.9% female) and similar body surface area (1.0 ± 0.4 m² vs 1.1 ± 0.4 m², p=0.74). S/p patients showed depressed TAPSE (17.2 ± 5.0 mm vs 21.1 ± 3.7 mm, p<0.0001) and tricuspid s’ (9.1 ± 2.0 cm/s vs 12.5 ± 2.1 cm/s, p<0.0001) in comparison with the controls. RV diastolic inflow pattern in the s/p group was characterized by lower tricuspid E wave (61.3 ± 14.4 cm/s vs 54.5 ± 8.7, p=0.020) and A wave (41.2 ± 14.0 cm/s vs 34.6 ± 7.9, p=0.019) as compared to controls, resulting in similar E/A ratio in the two groups (1.6 ± 0.5 vs 1.7 ± 0.5, p=0.69). However, patients in the s/p group showed lower tricuspid e' (12.2 ± 2.6 cm/s vs 15.3 ± 3.1 cm/s, p<0.0001) and higher tricuspid E/e' ratio (5.3 ± 1.9 vs 3.7 ± 0.7, p<0.0001) than the controls. An abnormal TAPSE (<16 mm) was present in 16 patients in the s/p group and in only one subject in the control group (43.2% vs 2.7%, p<0.0001). Similarly, reduced tricuspid s’ (<10 cm/s) was present in 26 patients in the s/p group and in 2 subjects in the control group (70.3% vs 5.4%, p<0.0001). A tricuspid E/e’ ratio >6, currently recommended as suggestive of pseudonormal RV filling and raised right atrial pressures, was found in 28 (75.7%) patients in the s/p group, and in none of the controls (p<0.0001).

Conclusions: Patients with previous surgical correction of congenital heart disease show significant RV systolic and diastolic dysfunction compared to matched controls. Considering the established prognostic importance of RV impairment, careful clinical and echocardiographic follow-up should be considered of clinical importance in these patients.
ROLE OF ECHOCARDIOGRAFI DURING NEONATAL PERIODE. FREQUENCIES AND TIME OF DIAGNOSIS OF CONGENITAL HEART SEPTAL DEFECTS (CHSD) IN NEONATAL PERIOD

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Septal defect (DIA, DIV, A-V defect) are the most frequent congenital heart defect, near 35-40% of all of them. Prevalence is 1-4/1000 live birth, and often are associated with other congenital heart defects. Large and not treated defects in the right moment are associated with serious complication like: heart failure, arrhythmia, pulmonary hypertension etc.

A systolic murmur can be the first sign of the congenital heart defect in a newborn babies, and in the same time the most habitual reason of for a cardiopediatric consultation. Prevalence of presence of a systolic murmur neonatal period is different in different articles and is published from 0.9 to 77.4%. This difference in prevalence seems to be related to with the size of the study.

Aim. To assess the frequency of congenital heart septal defect in neonate referred to our pediatric cardiology service, the single pediatric cardiac tertiary center in Albania. To asses the role of echocardiografi during neonatal period in our reality, when fetal echocardiografi is still imperfect.

Methods. Were analysed retrospectively the data-base of the outpatient clinic of our service, all neonates during june 2009-june 2012, referred for evaluation by the Pediatric community. Referral reasons was a systolic murmur. All patients were evaluated with physical examination and echocardiografi.

Results: A systolic murmur often is not associated with clinical manifestations, but is strongly related with a DIV. More affected are male neonates. The high incidence of these defects draws attention to a careful examination of newborns in terms of time to capture the heart defects that would later complicate the future of healthy children. Diagnosis and timely correction of congenital heart diseases, cures or improves quality of life.

Keywords: congenital heart diseases, septal defekt, DIA, DIV
CARDIAC FIBRO-FATTY RHABDOMYOMA IN AN ADULT PATIENT WITH TUBEROUS SCLEROSIS COMPLEX

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Introduction
Cardiac rhabdomyoma is the most common cardiac tumor in childhood (70-86%) frequently associated with Tuberous Sclerosis Complex (40-50% of cases), an autosomal dominant neurocutaneous disorder with multiorgan involvement.

Cardiac rhabdomyoma is usually the earliest hamartoma detected in TSC patients, with a peak of incidence in the first two years of life and tendency to spontaneous regression.

Cardiac involvement can range from small nodular lesions up to larger masses with potentially dangerous sequelae, such as obstruction of ventricular outflow tracts, impairment of ventricular filling or rhythm disorders.

Cardiac rhabdomyoma ultrasound features are round, homogeneous and hyperechoic intramyocardial masses, sometimes appearing as multiple foci.

Case report
We describe a fibro-fatty rhabdomyoma in a 23-years old boy with Tuberous Sclerosis Complex, characterized by multiorgan involvement: bilateral renal angiomyolipomas, multiple liver angiomyolipomas, subependymal astrocytoma, multiple benign brain tumors, cortical tubers, neck and shoulder angiofibromas, sacral osteolytic lesion. He is asymptomatic for chest pain, syncope or palpitations, referring good exercise tolerance. The electrocardiogram is normal. The echocardiogram shows three small hyperechogenic intraparietal masses in the interventricular septum, resembling rhabdomyomas. No impairment of ventricular filling or outflow tract obstruction is observed. No rhythm abnormalities has been found on 24-hours EKG Holter and stress test. Cardiac MRI confirms the presence of three small rhabdomyomas in the interventricular muscular septum with the signal intensity of fat and absence of enhancement.

Discussion
Cardiac rhabdomyoma is usually the earliest cardiac detectable hamartoma in TSC and tends to regress in the first years of life. Histopathologically, cardiac rhabdomyoma is composed of vacuolated myocytes. Although rhabdomyoma is well described in childhood, the pattern of the disease in adulthood is less known.

In this adult patient with TSC and multiorgan involvement, cardiac MRI has evidenced multiple ovoidal intramural homogeneous masses with imaging features compatible with fibro-fatty tissue. This finding, recently reported only in few case reports of adult patients with TSC, may suggest that cardiac rhabdomyoma can evolve to fibro-fatty tissue in the long term. In comparison with known causes of fat in the myocardium (epicardial fat deposit, arrhythmogenic right ventricular displasia, right ventricular fat, liposarcomas, etc) fibro-fatty masses in TSC seem to be characterized by well-circumscribed form, intramural location, pure fat density, absence of enhancement and of infiltrative features on surrounding tissue. Therefore, fibro-fatty mass in adult TSC patients may arise challenging diagnostic issues and need longer follow-up to define prognostic implications.