The Face Is the Mirror of the Soul. The Cardiovascular Physical Exam Is Not Yet Dead!

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Abstract: Cardiac pathology can be congenital or acquired with underlying genetic predispositions. In this era of medicine there is a concern that the comprehensive physical examination doctors prided themselves on is becoming a lost art. Research studies have also revealed a decline in physical examination skills. The full clinical cardiovascular examination is indeed quite complex and does take significant time to master. It is critical that physicians be competent in the physical exam. Not identifying subtle clinical findings leading to missed or delayed diagnosis which can lead to significant morbidity and mortality. In this paper we intend to highlight the clinical cardiovascular findings that may be detected on patients even before initiating the physical exam. The head and neck visual examination may be quite revealing. (Curr Probl Cardiol 2021;46:100644.)

Congestive Heart Failure

Heart failure incidence is rising due to the increasing age of the population as well as improved diagnostic techniques for the detection of heart failure.[1-4] Improved treatment and survival of patients with ischemic heart disease have also increased the incidence of heart failure.[3,4] Ischemic heart disease is the most common cause of heart

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failure. Chronic heart failure can cause body wasting, including temporal wasting and cachexia, signs that the disease has an extremely high mortality rate.[5] Cachexia is secondary to anorexia as a consequence of symptoms such as dyspnea and fatigue or due to interstitial edema causing nausea and protein-losing enteropathy. It is also associated with gastrointestinal fat malabsorption and increased resting metabolic rates in patients with increased heart failure severity.[6,7] The chronic sympathetic overdrive with increased inflammatory markers such as tumor necrosis factor-alpha is also associated with malnutrition and wasting.[8]

Acute and chronic heart failure is characterized by resting or exertional dyspnea. In the chronic stages, clinical signs of congestion become harder to detect as the lymphatic system becomes more efficient in drainage of the extravascular fluid. The bedside physical examination remains a critical component in the assessment and monitoring of acute heart failure patients, the cornerstone being evaluation of the Jugular Vein. In chronic heart failure, jugular vein distension (JVD) and peripheral edema predict poor prognosis.[9] The presence and extent of JVD is also a strong predictor of the severity of heart failure exacerbation. It is also an independently associated with both short- and long-term mortality.[10] JVD on cardiac exam correlates with elevated jugular venous pressure, which is a manifestation of abnormal right heart dynamics indicative of elevated pulmonary capillary wedge pressure from left heart failure.[11]

Exam Findings

The jugular vein should be assessed with the patient in a relaxed in a 45° incline position. The area of pulsatility in the neck should be closely evaluated. Differentiating venous from carotid arterial pulsations is a key component of this exam and based on the following points. The venous wave is bifid—it rises when you lower the head of the bed and sinks when you raise the head of the bed. It sinks into the chest with inspiration and is not palpable. Pressure on the right upper quadrant of the abdomen while watching the neck will help identify a rising venous pulsation.

Superior Vena Cava Syndrome

Superior Vena Cava (SVC) syndrome is caused by an obstruction of the SVC that impairs venous drainage from the head. The clinical findings in SVC syndrome are closely linked to venous congestion and the resultant elevation in venous pressures seen in the upper body. The patient will present clinically with facial and arm swelling. It is most commonly caused by

malignant mediastinal masses or pancoast tumor.[12] Horners syndrome may be seen in Pancoast tumors with the known triad of ptosis, myosis and anhidrosis which is also detected on the facial clinical exam. Increasingly it has been caused by indwelling intravascular devices such as catheters[13] as well as pacemakers and implantable cardioverter defibrillators leads.[14] These devices result in venous wall inflammation, fibrosis, and eventual thrombus leads to stenosis of the vessel itself. An estimated 15,000 cases of SVC syndrome occur each year in the United States, with studies pointing to increasing frequency due to the concomitant rise in the use of semipermanent intravascular catheters.[15] SVC syndrome is an emergency. It can raise intracranial pressure if the obstruction is severe, causing headache, dizziness, risk of aneurysm, or rupture of intracranial arteries. Management of the disease is guided by treating the patient's underlying SVC syndrome etiology. For patients with acute or subacute thrombus related to an indwelling intravascular device, removal should be considered along with anticoagulation therapy and catheter-directed thrombolysis or thrombectomy before venoplasty and stent placement. Multidisciplinary treatment planning for those with obstruction due to malignancy is essential, based on tumor type and staging for appropriate chemotherapy or radiation therapy.[16] With expanding treatment options for both benign and malignant etiology, endovascular therapy is now widely considered as the first-line treatment for SVC syndrome.[17]

Aortic Regurgitation

In the advanced stages aortic regurgitation can have very interesting head and neck findings. These include rhythmic head-bobbing (DeMusset's sign). The neck will reveal "dancing carotids." This is from a rapidly rising "water-hammer" pulse, which collapses suddenly as arterial pressure falls rapidly during late systole and diastole is seen in aortic regurgitation. This is known as Corrigan's pulse. Muller's sign is a bobbing of the Uvula in systole that is noticed in severe aortic regurgitation (AR).[18] Chronic AR is caused by disease of the valve leaflets or enlargement of the aortic root. In the developing world, the most common cause of AR is rheumatic heart disease. However, in developed countries, AR is most often due to aortic root dilation, congenital bicuspid aortic valve, endocarditis, and calcific valve disease.[19] The inability of the aortic valve leaflets to remain closed during diastole results in a portion of the left ventricular (LV) stroke volume leaking back from the aorta into the LV. The added volume of regurgitant blood produces an increase in LV end-diastolic volume and wall stress. The ventricle responds with

compensatory eccentric hypertrophy.[20] The heart initially adapts well to chronic AR, functioning as a very efficient and compliant high output pump. However, the increased stroke volume eventually results in distension of the peripheral arteries and an increase in systolic pressure. Regurgitation into the LV leads to a rapid fall in arterial pressure with a quick collapse of the arteries and a low diastolic pressure. The resulting wide pulse pressure leads to the characteristic physical findings in the head and neck. Additionally, chronic AR is associated with a distinctive early high-pitched diastolic "blowing" decrescendo murmur.[21] AR can progress to left heart failure if left untreated. Aortic valve surgery, predominantly aortic valve replacement, is the mainstay of treatment of symptomatic severe AR and some cases of asymptomatic AR.[22]

Tricuspid Regurgitation

In addition to congestive heart failure, SVC syndrome, and AR, several other cardiac problems cause JVD, and the neck exam for JVD is nuanced. Overall, the jugular venous pulsation can be categorized further into "A," "C," and "V" waveforms, each representing different phases of cardiac filling associated with different problems. Tricuspid regurgitation is characterized by the backflow of blood into the right atrium during systole. When TR is severe, right atrial and venous pressures rise and can result in the signs and symptoms of right-sided heart failure.[23] The cause of primary TR in adolescents and young adults usually is congenital, with Ebstein's anomaly being most common. Secondary TR is most commonly caused by pulmonary hypertension. There is a distinct "C-V" (regurgitant) wave on the JVD exam due to systolic regurgitation into the right atrium.[24] The jugular vein is very pulsatile and may be confused with the carotid arterial pulse. Jugular venous distension may be more prominent with inspiration (Kussmaul's sign), a result of the increase in venous return. A systolic thrill may be felt over the jugular vein in patients with severe regurgitation. Diuretics, especially loop diuretics, are needed to treat volume overload in patients with severe TR and right-sided heart failure. Treatment of a patient's pulmonary hypertension may also improve TR.[25] Tricuspid valve repair is generally preferred to tricuspid valve replacement, with valve replacement performed only when a repair is not feasible.[26]

Ventricular Tachycardia

In ventricular tachycardia (VT) patients are usually in critical condition. It is not uncommon for patients to present with "stable VT." The diagnosis

of a stable wide complex rhythm is indeed a challenge, and the physical exam may also play a role. The neck veins show Cannon "A" waves, when the right atrium is contracting against a closed tricuspid valve due to asynchronous atrioventricular dissociation, a hall mark of VT. "A" waves are intermittent and irregular jugular venous pulsations of greater amplitude than regular waves.[27] They reflect simultaneous atrial and ventricular activation, resulting in contraction of the right atrium against a closed tricuspid valve. Prominent A waves can also be seen during some SVTs. Such prominent waves result from simultaneous atrial and ventricular contraction occurring with every beat. Synchronized cardioversion remains the main therapy in a hemodynamically unstable patient.[28]

Hypercholesterolemia

Xanthomas are well-circumscribed flat lesions in the connective tissues of the skin and fascia that mainly consist of foam cells formed from macrophages as a result of excessive uptake of low-density lipoprotein (LDL) particles. Patients with hypercholesterolemia, in particular familial hypercholesterolemia (FH), have a predisposition to high accumulation of the lipid particles in the vascular walls. The accumulation of excessive lipid particles causes the development of premature atherosclerosis.[29] The most common clinical manifestation of FH is tendinous xanthomas in Achilles tendons and tendons of the extensors such as the back of the hands. Xanthelasmas are straw to orange-yellow colored cholesterol-rich deposits that usually present as bilateral, symmetrical, soft, yellow papules over the eyelids and around the corneal arcus. Arcus corneae is a white-yellowish lipid-laden deposit located near the periphery of the cornea. Xanthelasmas are usually diagnosed by clinical appearance but are highly suspicious of an underlying disease. Studies have shown that xanthelasmas are associated with increased atherosclerotic cardiovascular disease and reduced lifespan.[30,31] Xanthelasma in hyperlipidemic patients are associated with lower hyperlipidemia and higher LDL levels, both of which are established atherogenic risk factors. Carotid intima-media thickness measurement has been used as a surrogate marker for generalized atherosclerosis and risk of cardiovascular disease. Studies have found an increase in mean Carotid intima-media thickness values in patients with xanthelasmas.[32,33] Facial xanthomas are prevalent in FH with very high LDL cholesterol levels and are often the initial clinical manifestation of the acute coronary syndrome.[34] Xanthelasmas are associated with an increased risk of ischemic heart disease, myocardial infarction, and early mortality independent of known cardiovascular risk factors such as plasma cholesterol and

triglyceride concentrations.[35] Arcus corneae is seen in patients with xanthelasma, but it is not considered an independent predictor of cardiovascular risk and increased mortality.

Lyme Disease

Lyme disease is the most common vector-borne disease in the United States, and it involves multiple body systems. It is caused by infection with Borrelia burgdorferi. It typically begins with a unique skin lesion, erythema chronicum migrans. Craniofacial and cardiac manifestations occur a few weeks to months after the initial skin lesion. Neurological involvement occurs in 12%-14% of Lyme disease patients, and approximately half of these involve cranial nerve palsy. Lyme disease is one of the most common causes of seventh nerve paralysis.[36] Patients with seventh nerve paralysis present with ipsilateral inability to elevate the eyebrow, ptosis, drooling, and hyperacusis. Lyme carditis is a rare manifestation of Lyme disease and can present with symptoms such as chest pain, shortness of breath, palpitations, and syncope. Cardiac involvement can include atrioventricular block as well as myocarditis with or without pericardial involvement.[37] Third-degree heart block is the most common manifestation, presenting in approximately 49% of patients. Patients with myocardial involvement can present with episodes of nonsustained VT, ST-segment depression, and congestive heart failure.[38] Less common cardiac manifestations of Lyme disease include pericardial effusion, myocardial infarction, and QT interval prolongation. Valvular involvement, including Lyme endocarditis, is a rare manifestation of lyme disease.[39]

Wilson Disease

Wilson disease (WD) is a copper deposition disease that most classically affects the liver and brain. The classic presentation of WD is jaundice and copper deposition in the cornea known as Kayser−Fleischer rings. It has an autosomal recessive inheritance and classically presents in patients between 5 and 35 years of age.[40] Patients commonly develop acute hepatitis and acute liver failure, with Coombs negative hemolytic anemia. Neurologic symptoms include dysarthria, gait abnormalities, ataxia, dystonia, tremor, and Parkinsonism. Patients may experience depression, personality changes, irritability, and impulsiveness. WD affects most of the major organs, including the heart. Studies have shown that WD is associated with increased thickness of the interventricular septum and LV posterior wall.[41] Concentric LV remodeling and LV hypertrophy have also been shown.[42]

Electrocardiogram shows a relatively high frequency of benign supraventricular tachycardias and extrasystolic beats. Signs of structural heart disease in WD have not been found.[43] WD is treated with D-penicillamine, Trientine, or Zinc.[44] The prognosis in patients with WD is excellent except those with advanced disease and those presenting with rapidly progressive liver failure and hemolysis. The neurologic, psychiatric, and hepatic abnormalities gradually improve with treatment, and the results usually return to normal liver biochemical tests.

Lupus

Systemic lupus erythematosus (SLE) is a complex autoimmune disease with the predilection of multiple organ systems in the body. Cutaneous and articular manifestations are the most common signs in SLE. Cutaneous manifestations can be acute, subacute, or chronic.[45] The most common specific facial manifestation of the acute cutaneous lupus is a malar eruption or "butterfly" rash that involves the cheeks but spares the nasolabial folds. The most common cardiac side effect of SLE is pericarditis. Another very important side effect is Libman−Sacks endocarditis. This is a form of nonbacterial endocarditis specific to SLE and one of its most common heart-related manifestations of SLE.

SLE is also associated with accelerated atherosclerosis and increased cardiovascular complications. Premature atherosclerosis is the most frequent cause of coronary artery disease and ischemic heart disease in SLE. Evidence has demonstrated that extensive vascular damage has already occurred before the clinically overt cardiovascular disease has developed in lupus patients.[46] The most common underlying etiologies of acute myocardial infarction in SLE patients include coronary atherosclerosis, arteritis, myocarditis, and thrombosis. Some cardiovascular abnormalities are associated with positive anti-Ro/SSA, anti-La/SSB, anticardiolipin, anti-dsDNA antibodies, which are highly associated with certain facial dermatological lupus manifestations, especially discoid lupus.[47] Elevated inflammatory markers in the presence of active facial skin lesions are highly associated with cardiovascular disease in SLE.

Myotonic Dystrophy

Myotonic dystrophy (MD) is a multisystem autosomal dominant disease characterized by myotonia, progressive muscle weakness, and distal atrophy of the extremities. Myotonic dystrophy can present with several orofacial abnormalities such as atrophy and degeneration of masticatory muscle.[48,49] The facial appearance of MD patients is characteristic of the

disease and easily identifiable. The classical "facies" is caused by weakness and wasting of the facial, masticatory, and levator palpebral muscles. Affected individuals can present with ptosis, and the typical myopathic or "hatched" appearance. MD can also present with frontal baldness and lenticular opacities. Other facial manifestations can include temporomandibular joint disorders that can present with joint clicking and locking resulting in impaired chewing function and decreased maximum bite force. Rarely, there can be cutaneous involvement of the face presenting clinically with alopecia, dysplastic nevi seborrheic dermatitis, and oral hemangiomas.[50]

The clinical manifestation of MD on the heart include conduction system abnormalities. Fatty infiltration and cardiac fibrosis can affect the entire conduction system from the sinoatrial and atrioventricular nodes to the His-Purkinje bundle. This can result in ectopic activity and re-entrant arrhythmias which leads to supraventricular arrhythmias, ventricular arrhythmias and intraventricular conduction defects.[51] Heart failure can present with LV systolic or diastolic dysfunction.[52,53] Sudden cardiac death is the most frequent cardiovascular cause of death in patients with MD which may be related to the ventricular arrhythmias.

Marfan Syndrome

Marfan syndrome (MFS) is a connective tissue disorder of variable inheritance that affects multiple organ systems with cardiovascular, ocular, and skeletal abnormalities. Skeletal abnormalities can include an excessive length of upper and lower extremities, hyperlaxity, and scoliosis. Craniofacial manifestations can present clinically with temporomandibular disorders, maxillary and mandibular prognathism, long narrow teeth, hypodontia, high-arched palate, and dislocation of the lens. Periodontal disease occurs with a higher frequency and severity among maxillofacial disorders in MFS. Clinically patients with MFS have a narrow cranium and retrognathia or micrognathia of the upper jaw. Maxillary bone is hypoplastic and causes dental crowding. Teeth consist of hypoplastic enamel, and there is an increased incidence of dental carriers and severe periodontal disease.[54] Cardiovascular complications constitute the main cause of mortality in individuals affected with MFS. The most common cardiovascular manifestation is the dilation of the ascending aorta associated with life-threatening aortic dissection.[55,56] An aortic aneurysm is also common in MFS and has a high risk of aortic dissection. Individuals with MFS have a higher incidence of multiple abnormal cardiac valves with prolapse and regurgitation. In MFS, severe myxomatous

mitral valve prolapse can often coexist with aortic root aneurysm. Pulmonary artery root may be affected by the same connective tissue defect as the aortic root in MFS. The dilation of the main pulmonary artery is highly prevalent in individuals with MFS.[57] The involvement of valves also predisposes these individuals to a higher risk of contracting bacterial infective endocarditis.[58] Previous studies have revealed a strong relationship between periodontal disease and cardiovascular complications in individuals with MFS.[59] Heart failure is the second leading cause of death in MFS. Mitral valve regurgitation in the setting of mitral valve prolapse and aortic valve regurgitation in the setting of aortic root dilation is the cause of heart failure in MFS.[60]

Down Syndrome, Edwards Syndrome, and Patau Syndrome

Down syndrome is a chromosomal genetic disorder causing developmental and intellectual delays. Down syndrome is associated with an atrioventricular septal defect, a severe heart abnormality. It is also associated with Tetralogy of Fallot and persistent Ductus Arteriosus.[61] Facial features associated with Down syndrome include slanting eyes, small chin, round face, flat nasal bridge, brushfield spots in the iris, irregular outer ears, and a flattened nose.[62]

Edwards syndrome is a genetic chromosome disorder, also known as Trisomy 18, in which most individuals affected do not survive the age of one. Those that do survive their first year tend to have health-related complications and lying in those complications; congenital heart diseases are quite common. The prominent craniofacial abnormalities associated with Edwards Syndrome are microretrognatia, dysplastic ears, prominent occipital, retroverted ears, low-set ears, and blepharophimosis/short palpebral fissures. Another relevant abnormality, though less frequent, includes cleft lip, bilateral cleft lip, and palate cleft. Some unusual manifestations can include microtia, preauricular, facial paralysis, encephalocele, and lack of external auditory canal.[63] Ventricular septal defect, atrial septic defect, and patent ductus arteriosus are examples of congenital heart diseases that may be present in patients with Edwards syndrome.[64]

Patau syndrome, or Trisomy 13, is another chromosomal genetic disorder. Similar to Edwards syndrome, the life span is relatively short for Patau patients, and most patients who live past the age of 1 year tend to have an increased susceptibility to cardiovascular diseases. Facial defects include cyclopia, cleft lip, cleft palate, a sloping forehead, deformed ears, anophthalmia or microphthalmia, micrognathia, and preauricular tags.

The spectrum of cardiac disease in Patau syndrome includes a ventricular septal defect, atrial septal defect, tetralogy of Fallot, atrioventricular septal defect, and double outlet right ventricle.[65]

Noonan Syndrome

People with Noonan syndrome have distinctive facial features such as deep grooves in the area between the nose and mouth, widely spaced eyes that are usually pale-blue or blue-green, and low-set of ears that are rotated backward. Cardiac manifestations associated with Noonan syndrome include pulmonary stenosis, hypertrophic cardiomyopathy, Secundum atrial septal defect, ventricular septal defect, peripheral pulmonary stenosis, atrioventricular canal, aortic stenosis, mitral valve abnormalities, aortic coarctation, and coronary artery anomalies. Hypertrophic cardiomyopathy can be mild or severe and can present from the prenatal period to late childhood. Nearly 25% of patients die because of heart failure in the first year. Those who live past their first year tend to face detrimental cardiovascular complications.[66]

Williams Syndrome

Williams syndrome is a chromosomal genetic disorder that manifests as mental retardation and presents with distinctive facial features. Facial features can include a small upturned nose, long philtrum, wide mouth, full lips, small chin, and puffiness around the eyes.[67] Cardiovascular abnormalities occur in 80% of Williams syndrome patients, with the majority of these due to arterial stenoses.[68]

Cyanotic Congenital Heart Disease

Cyanotic congenital heart disease encompasses heart defects present at birth and subsequently results in a low blood oxygen level. These diseases include the 5 Ts: tetralogy of Fallot, transposition of the great arteries (TGA), total anomalous pulmonary venous return, truncus arteriosus, and tricuspid atresia. All 5 of these diseases can assist the physician in reading their patients' hearts from their faces. Tetralogy of Fallot is a rare condition but is the most common of the 5 Ts. It is caused by a combination of 4 heart defects that are present at birth. Tetralogy of Fallot defects causes oxygen-poor blood to flow out of the heart and into the rest of the body. Dysmorphic facial features are present in 9 of 31 children with tetralogy of Fallot (29%). These irregularities include hypertelorism, low-set ears, small mouth, short philtrum, and micrognathia.[69] TGA is a complex congenital heart defect. In a normal heart, 2 large arteries carry blood out of the heart. In children with TGA, these 2 arteries leaving the

heart are reversed. People who have TGA have abnormal facial features manifesting clinically as a blue color of the skin and cachectic appearance due to poor weight gain.[70] Total anomalous pulmonary venous return is a heart disease in which the 4 veins that take blood from the lungs to the heart do not normally attach to the left atrium. Instead, they attach to another blood vessel or the wrong part of the heart. Patients with Total anomalous pulmonary venous return can be identified clinically by a broad forehead, periorbital fullness, flattened nasal bridge with an upturned nose, long philtrum, and rounded cheeks. Truncus arteriosus is a rare type of heart disease in which a single blood vessel (truncus arteriosus) comes out of the right and left ventricles rather than 2 vessels (pulmonary artery and aorta). People with truncus arteriosus have facial features that include hypertelorism, low-set ears, micrognathia, down-slanting palpebral fissures, short philtrum, and chromosome 22q11 deletion, and small mouth.[71] Tricuspid atresia is a type of heart disease present at birth (congenital heart disease) in which the tricuspid heart valve is missing or abnormally developed. The defect blocks blood flow from the right atrium to the right ventricle. People with tricuspid atresia have facial anomalies such as bluish skin color and smaller ears and nose due to reduced growth.

DiGeorge Syndrome

DiGeorge syndrome, also known as Velocardiofacial syndrome, is considered a primary immunodeficiency that is characterized by cellular (T-cell) deficiency, characteristic facies, congenital heart disease, and hypocalcemia. DiGeorge syndrome is caused by abnormal formation of certain tissues during fetal development. This deletion syndrome is widespread and affects nearly 1 in 3000 children. Common facial characteristics of patients with DiGeorge syndrome include small and low-set ears, short width of eye openings, hooded eyes, relatively long face, enlarged nose tip, and a flattened groove in the upper lip. The most common facial characteristics in children are stated above but are predominantly cleft lip and palate.[72]

Conclusion

The physical examination is an integral part of the cardiovascular system evaluation. There is a wide array of physical findings on the head and neck of such patients. Prompt identification of such findings may limit the diagnostic tests required. What you do not actively look for in a physical examination you will most likely miss. Through a holistic knowledge of disease processes and practicing the art of the physical exam, clinicians can "see" anatomy and hemodynamics of a patient's heart through their face.

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