Percutaneous trans-luminal management of the vascular manifestations of neurofibromatosis type 1

Poster No.: C-0312
Congress: ECR 2018
Type: Educational Exhibit
Authors: J. Raborn\textsuperscript{1}, A. Gunn\textsuperscript{1}, K. Mahmoud\textsuperscript{1}, S. Moawad\textsuperscript{1}, M. Poundstone\textsuperscript{1}, A. M. K. Abdel Aal\textsuperscript{1}, S. Saddekni\textsuperscript{2}; \textsuperscript{1}Birmingham/US, \textsuperscript{2}Birmingham, AL/US

Keywords: Congenital, Education, Arterial access, Angioplasty, CT-Angiography, Vascular, Soft tissues / Skin, Arteries / Aorta, Genetic defects

DOI: 10.1594/ecr2018/C-0312

Any information contained in this pdf file is automatically generated from digital material submitted to EPOS by third parties in the form of scientific presentations. References to any names, marks, products, or services of third parties or hypertext links to third-party sites or information are provided solely as a convenience to you and do not in any way constitute or imply ECR's endorsement, sponsorship or recommendation of the third party, information, product or service. ECR is not responsible for the content of these pages and does not make any representations regarding the content or accuracy of material in this file.

As per copyright regulations, any unauthorised use of the material or parts thereof as well as commercial reproduction or multiple distribution by any traditional or electronically based reproduction/publication method is strictly prohibited.

You agree to defend, indemnify, and hold ECR harmless from and against any and all claims, damages, costs, and expenses, including attorneys' fees, arising from or related to your use of these pages.

Please note: Links to movies, ppt slideshows and any other multimedia files are not available in the pdf version of presentations.

www.myESR.org
Learning objectives

1. Review the clinical manifestations of Neurofibromatosis type 1 (NF1).

2. Discuss the vascular pathology associated with NF1.

3. Review the clinical presentation, diagnosis, and percutaneous management of NF1 related renal artery stenosis, middle aortic syndrome, and cerebral artery disease.
Background

Neurofibromatosis 1 (NF1) is an autosomal dominant disorder found in approximately 1 out of every 3,000 individuals. The disorder results from a microdeletion in the \textit{NF1} gene on chromosome 17q11.2 that results in premature translation termination of the neurofibromin protein. The classic clinical presentation includes cutaneous café au lait spots, plexiform neurofibromas, pigmented hamartomas of the iris ("Lisch nodules"), and freckling of the axillary or inguinal regions. Less common clinical presentations can also include: optic gliomas, macrocephaly, scoliosis, muscle weakness, hemi-hypertrophy, long bone dysplasia, pseudo-arthritis, epilepsy, and behavior and/or psychological disorders. Yet, given that expression of NF1 is variable, patients can present with many or few of these features. Therefore, diagnosis relies on criteria established by the National Institutes of Health (NIH) requiring the presence of at least two of their seven diagnostic criteria (#6 café au lait spots, #2 neurofibromas, axillary freckling, #2 Lisch nodules, sphenoid dysplasia, thinning of a long bone cortex, or a first-degree relative with NF1). Given the complexity of the disorder, longitudinal surveillance of patients with NF1 for complications or new manifestations is needed.

NF1 can have vascular manifestations as well. NF1 vasculopathy includes aneurysms, stenosis, and arterio-venous malformations (AVMs). The exact incidence of vascular involvement is unknown but is estimated at somewhere between 0.4-6.4% of patients with NF1. Although, the incidence could be significantly higher since approximately 50% of patients with NF1 vasculopathy are asymptomatic and vascular involvement is found incidentally. The most common clinical presentation of NF1 vasculopathy is hypertension secondary to renal artery stenosis or aortic coarctation. Additionally, patients can present with vessel rupture, thromboembolism from aneurysms, neurologic impairment due to carotid involvement or nerve impingement, abdominal pain from visceral artery involvement, or claudication secondary to involvement of the arteries to the extremities. Vasculopathy is the second-leading cause of death in patients with NF1; thus, its recognition and management is an important concept. The purpose of this poster will be to outline the most common presentations of NF1 vasculopathy, discuss the technical approach to the endovascular management of NF1, and review their clinical outcomes.
Findings and procedure details

Renal arterial involvement

NF1 vasculopathy most commonly affects the renal artery. Renal artery stenosis (RAS) is a more common presentation than renal artery aneurysms. Patients with RAS typically present with hypertension and/or renal dysfunction. The initial diagnostic modality of choice is ultrasound, which can show broadened spectral waveforms, increased peak systolic velocity, and parvus et tardus waveforms distal to the stenosis. Treatments are aimed at improving renal perfusion through percutaneous transluminal angioplasty (PTA) of the renal arteries (with or without stent placement) and/or surgical intervention. Medical management includes inhibiting the renin-angiotensin system with angiotensin converting enzyme inhibitors or angiotensin II receptor blockers. Pre-procedure planning is done with dedicated CT or MR angiography (Fig. 1 on page 6). PTA is often the first line therapy for RAS in NF1 (Fig. 1 on page 6, Fig. 2 on page 6, Fig. 3 on page 7, and Fig. 4 on page 8). PTA has technical success rates up to 72% and clinical success in reducing blood pressure in up to 39-59% of patients. Yet, re-stenosis occurs in about 44% of patients. Some patients may require surgical re-vascularization (which has ~80% clinical success rate) but PTA has a reduced morbidity and shorter hospital stay, and should thus be considered prior to surgery. The long-term results of PTA for RAS in NF1 vasculopathy have not been documented.

Aortic and mesenteric artery involvement

Coarctation of the abdominal aorta occurs in approximately 12% of patients with NF1 and has been referred to as Middle Aortic Syndrome (MAS). This is commonly associated with stenosis of the mesenteric arteries (33%) as well as in conjunction with RAS. Aneurysms of the abdominal aorta can also be seen, although this occurs much less frequently. Patients with MAS usually present with intermittent abdominal pain and/or leg claudication. Screening ultrasound usually reveals narrowing of the abdominal aorta; however, CTA is usually obtained to determine the extent of disease (Fig. 2 on page 6). Indications for invasive management in patients with MAS are not defined but intervention should be considered when patients are symptomatic. A multidisciplinary approach is often required due to variation in stenosis location, segment length, number of lesions, co-morbidities, and response to medical therapy. PTA with or without stent placement is primarily performed for stenoses of mesenteric and renal arteries (Fig. 5 on page 9 and Fig. 6 on page 10). PTA of the abdominal aorta is generally not performed. Treatment for severe, symptomatic aortic coarctation typically includes endovascular stent placement or surgical repair. Endovascular and surgical success rates are not reported in this patient population but complication and mortality rates favor first-line endovascular repair (13% vs. 9% and 2.7% vs. 2.3%, respectively).
Carotid, vertebral, and cerebral artery involvement

Cerebrovascular manifestations occur in approximately 19% of NF1 patients and typically involve aneurysms of the internal carotid artery and its major branches or stenosis/occlusion of more distal intracranial arteries. Aneurysm of the internal carotid artery or major branches is most common and usually presents with or without headache and/or other neurologic deficit. These lesions are usually treated with endovascular coil placement or surgical clip placement. Other less common manifestations (AVF, hemorrhage, stenosis, etc) are treated similar to the general population with these disorders.
Fig. 1: A 4 year-old female with NF-1 presented with uncontrolled hypertension (170/120) and headaches, already taking Enapril. A, Coronal 3D reconstruction from an abdominal CTA shows ostial narrowing of bilateral renal arteries (blue arrows). B, Digital subtraction angiogram (DSA) of the aorta in a coronal projection confirms the CTA findings of bilateral renal artery stenosis (blue arrows) with post-stenotic dilatation. C and D, Frontal angiography images demonstrating balloon angioplasty of the left and right proximal renal arteries at the ostial stenoses with a 3.5 mm balloon (red arrows) over a 0.018” wire (blue arrow). E, Post-angioplasty DSA aortogram in a frontal projection shows mild residual stenosis of both renal arteries (blue arrows) with significantly improved flow.

She tolerated the procedure well and her blood pressure at 6 month follow-up was 115/60.

© University of Alabama at Birmingham (UAB)
**Fig. 2:** 8 year-old female with NF-1 who presented with uncontrolled hypertension. A, Coronal 3D reconstruction from an abdominal CTA shows mild narrowing of the infra-renal aorta (white arrow) and bilateral renal artery stenosis (blue arrows). Note the delayed enhancement of the lower pole of the right kidney (yellow arrow) and collateral vessels (red arrow). B, DSA aortogram confirms the presence of a narrowed infra-renal aorta (white arrow) and bilateral renal artery stenosis (blue arrows) with post-stenotic dilatation. Note the collateral flow (red arrows) arising from the aorta, external iliacs, and lumbar arteries to fill both renal arteries. C, Later phase from the same aortogram as “B” more clearly shows the collateral vessels (red arrows). D, Even later phase from the same aortogram as “B” and “C” shows the flow into the right renal artery from collateral vessels (red arrow).

© University of Alabama at Birmingham (UAB)
Figure 3. 8 year-old female with NF-1 who presented with uncontrolled hypertension. A, Balloon angioplasty of the right renal artery stenosis was performed with a 5mm balloon (blue arrow) over an 0.018” wire (red arrow). B, Post-angioplasty DSA of the right renal artery shows an excellent response to treatment (blue arrow).

Fig. 3: 8 year-old female with NF-1 who presented with uncontrolled hypertension. A, Balloon angioplasty of the right renal artery stenosis was performed with a 5mm balloon (blue arrow) over an 0.018” wire (red arrow). B, Post-angioplasty DSA of the right renal artery shows an excellent response to treatment (blue arrow).

© University of Alabama at Birmingham (UAB)
Fig. 4. 8 year-old female with NF-1 who presented with uncontrolled hypertension. A, Balloon angioplasty of the left renal artery stenosis was performed with a 5mm balloon (blue arrow) over an 0.018” wire (red arrow). B, Post-angioplasty DSA of the left renal artery shows an excellent response to treatment (blue arrow).

She responded well clinically to treatment with a discharge blood pressure of 110/65.

**Fig. 4:** 8 year-old female with NF-1 who presented with uncontrolled hypertension. A, Balloon angioplasty of the left renal artery stenosis was performed with a 5mm balloon (blue arrow) over an 0.018” wire (red arrow). B, Post-angioplasty DSA of the left renal artery shows an excellent response to treatment (blue arrow). She responded well clinically to treatment with a discharge blood pressure of 110/65 mm Hg.

© University of Alabama at Birmingham (UAB)
Fig. 5: 38 year-old male with NF-1 who presented with post-prandial abdominal pain. A, Sagittal reformat from an abdominal CTA shows mild stenosis of the celiac artery (red arrow) and severe stenosis of the superior mesenteric artery (SMA) (blue arrow). Each artery shows post-stenotic dilatation. B, Coronal reformat from an abdominal CTA shows robust collaterals (blue arrows) from the inferior mesenteric artery (IMA) feeding the SMA territory. C, Digital subtraction angiogram (DSA) of the aorta in a lateral projection confirms the CTA findings of celiac (red arrow) and SMA stenosis (blue arrow). Note the robust size of the IMA (white arrow). D, DSA aortogram in a frontal projection again shows prominent collateral vessels from the IMA supplying the SMA territory (blue arrows).

© University of Alabama at Birmingham (UAB)
Fig. 6: 38 year-old male with NF-1 who presented with post-prandial abdominal pain. A, Sagittal DSA of the SMA with the tip of the sheath (red arrow) at the SMA origin and a diagnostic catheter (blue arrow) past the area of stenosis (white arrow). B, Balloon angioplasty was performed with a 5mm balloon (blue arrow) over an 0.018” wire (red arrow). C, Post-angioplasty DSA of the SMA in a lateral projection shows only mild improvement in the stenosis (white arrow). The decision was then made to stent the lesion. D, Post-stenting DSA of the SMA in a lateral projection show improvement in the stenosis after placement of a 7mm balloon-expandable stent (white arrow). The patient’s post-prandial pain improved significantly after this intervention and remained controlled for 7 years.

© University of Alabama at Birmingham (UAB)
Conclusion

The management of NF1 vasculopathy involves a combination of medical therapy with either surgical or percutaneous interventions. Specific indications for intervention are poorly defined; however, the overall goal is symptom alleviation. Appropriate patient selection based on the degree of vascular involvement, co-morbidities, and lesion morphology can improve outcomes. Complications, restenosis, and clinical failure are not uncommon even with improvements in surgical and percutaneous management.
Personal information

Ahmed Kamel Abdel Aal, MD, PhD, FSIR
University of Alabama at Birmingham (UAB).
Department of Radiology
Division of Interventional Radiology. United States, Alabama.
Address: 619 19th Street South, Birmingham, AL 35249.
Tel: (205) 975-4850
Fax: (205) 975-5257.
Email: akamel@uabmc.edu
Twitter: ahmed_kamel_ir
References


