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RARE DISEASES IN THE SLOVAK REPUBLIC
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PULMONARY ARTERIAL HYPERTENSION CENTRE – A MODEL FOR RARE DISEASE MANAGEMENT

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Pulmonary arterial hypertension (PAH) is a rare disease with average median survival rate about 3 years from the establishment of the diagnosis, except for PAH associated with congenital heart diseases. Diagnosis and management of PAH concentrate in the dedicated centres (reference centres, centres of expertise). The purpose of a reference centre is to undertake assessment and investigation of all causes of pulmonary hypertension, PAH-specific drug therapy, cooperation with other healthcare specialist, and to undertake research and education. In general, high volume centres achieve best outcomes, because of effective concentration of the specific experience and skills needed for rare disease management.
The paper brings brief characteristic of the reference centre for PAH according the guideline of European Society of Cardiology as well as own experience in this field.

Keywords: *pulmonary arterial hypertension – managment – reference centre*

Pulmonary arterial hypertension (PAH) is a rare but life-threatening disease. Median survival rate varies depending on PAH cause, and is on average about 3 years from the establishment of the diagnosis, except for PAH associated with congenital heart diseases. Incidence and prevalence have not been fully understood yet. Estimated prevalence of all types of PAH together is about 15-50 cases per million. The prevalence of PAH in certain at-risk groups is substantially higher. For example, in HIV-infected patients the prevalence is 0.5%, in patients with systemic sclerosis it has been reported to be 7-12%, and in patients with sickle cell disease the prevalence is around 2-3.75%.

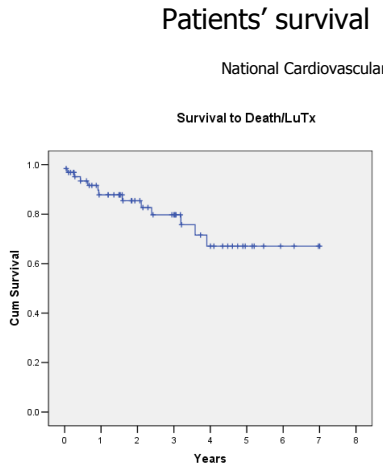
Idiopathic PAH (IPAH) has an annual incidence of 2 cases per million people and is 2-4 times as common in women as in men. The mean age at diagnosis is around 45 years, although the onset of symptoms can occur at any age.

Diagnosis and management of PAH concentrate in the dedicated centres. The purpose of a reference centre (centre of expertise) is to undertake assessment and investigation of all causes of pulmonary hypertension, PAH-specific drug therapy, work with other healthcare providers to obtain best outcomes for patients, and to undertake research and education. In general, high volume centres achieve best outcomes, because of effective concentration of the specific experience and skills needed for rare disease management.

The centre will need to have sufficient patients on chronic therapy and also new referrals to sustain high quality patient care. According to the Guidelines of European Society of Cardiology, a reference centre should follow at least 50 patients with PAH or chronic thromboembolic pulmonary hypertension (CTEPH) and should receive at least two new referrals with documented PAH or CTEPH per month.

Reference centres have to provide care by a multi-professional team, which should be comprised at minimum of two certified physicians (normally either or both with cardiology and respiratory medicine practice) with a special interest in PAH, clinical nurse specialist, radiologist with expertise in PAH imaging, access to psychological and social work support, and on-call cover. PAH centres have to be equipped with a ward where staff have special expertise in PAH, an intensive therapy unit, a specialist outpatient service, diagnostic investigations including echocardiography, CT scanning, nuclear scanning, magnetic resonance imaging, ultrasound, exercise testing, lung function testing, and catheterization laboratory (with expertise in performing vasoreactivity test) and access to the full range of specific PAH drug therapy available in the country. There should also be clearly established links to other services, which may not necessarily be on the same site. These links include services for connective tissue disease service, pulmonary endarterectomy, lung transplantation, adult congenital heart disease and genetics. Reference centres are required to undertake, register and provide patient survival analysis. It is expected that PAH centres will participate in collaborative clinical research in PAH, which includes phase II and phase III clinical trials and grant regular education about all clinical aspects of PAH to appropriate healthcare professionals. Cooperation with the national and/or European pulmonary hypertension patients association is also an important part of centre's activity.

Between Sept 1st 2005 and June 30th 2011, there were 129 patients with PAH identified and treated in Slovakia. Nowadays, PAH patients' health care in Slovakia is concentrated to five centres allowed to prescribe PAH specific drugs − four for adults and one for children with PAH (In comparison there are 3 PAH centres in Czech republic, 9 in Great Brittan or 3 in Portugal). The major one affiliated to the Department of heart failure/transplantation in National Cardiovascular Institute has cared for 90 PAH patient. The department is fully equipped by trained staff and all diagnostic techniques meet the strict criteria of ESC of PAH centre. Lung transplantation and endarterectomy are performed abroad in Vienna and Prague, respectively. Survival of 64 incidental PAH cases diagnosed in the centre in Figure 1.



Time Elapsed (years)	pts. survived (%)	95% CI (%)
1	88	84-92
2	85	80-90
3	76	69-83
4	67	59-75

Figure 1 Patients' survival in incident PAH

CONCLUSION

Medical care of patients with PAH is well established in Slovakia. Almost all drugs for PAH treatment registered in Europe are available. The presence of four PAH adult centres for population of 5.5 million inhabitants appears to be ineffective and might threaten the centres' level of expertise.

REFERENCES

National Pulmonary Hypertension Centres of the UK and Ireland. Consensus statement on the management of pulmonary hypertension in clinical practice in the UK and Ireland. Heart 94, 2008:1-41.

Galie N, Hoeper MM, Humbert M. et al. Guidelines for the diagnosis and treatment of pulmonary hypertension. Eur Heart J. 30, 20, 2009:2493-2537.

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CENTRUM PRE PĽÚCNU ARTÉRIOVÚ HYPERTENZIU – MODEL V MANAŽMENTE ZRIEDKAVÝCH CHORÔB

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Pľúcna artériová hypertenzia (PAH) je zriedkavé ale život ohrozujúce ochorenie. Medián prežívania je variabilný v závislosti od príčiny PAH. V priemere sa odhaduje na 3 roky od stanovenia diagnózy, okrem situácií kedy PAH vznikla v súvislosti s vrodenou srdcovou chybou. Diagnostika a manažment PAH sa koncentrujú v špeciálne určených centrách. Úlohou centra je zabezpečiť diagnostiku a starostlivosť o všetky typy PAH. Centrum poskytuje špecifickú liečbu, spolupracuje s odborníkmi z ostatných oblastí a je aktívne v oblasti vedy a výskumu. Tiež podporuje a edukuje pacientov formou spolupráce s pacientskymi organizáciami.
Vo všeobecnosti, výsledky centra súvisia s jeho vyťaženosťou. Centrá s vyššou vyťaženosťou dosahujú najlepšie výsledky a to najmä vďaka pravidelnému tréningu špecifických zručností a odborných skúseností.
Práca prináša stručnú charakteristiku centra pre PAH podľa odporúčaní Európskej kardiologickej spoločnosti ako aj vlastné skúsenosti Národného ústavu srdcových a cievnych chorôb v Bratislave.

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