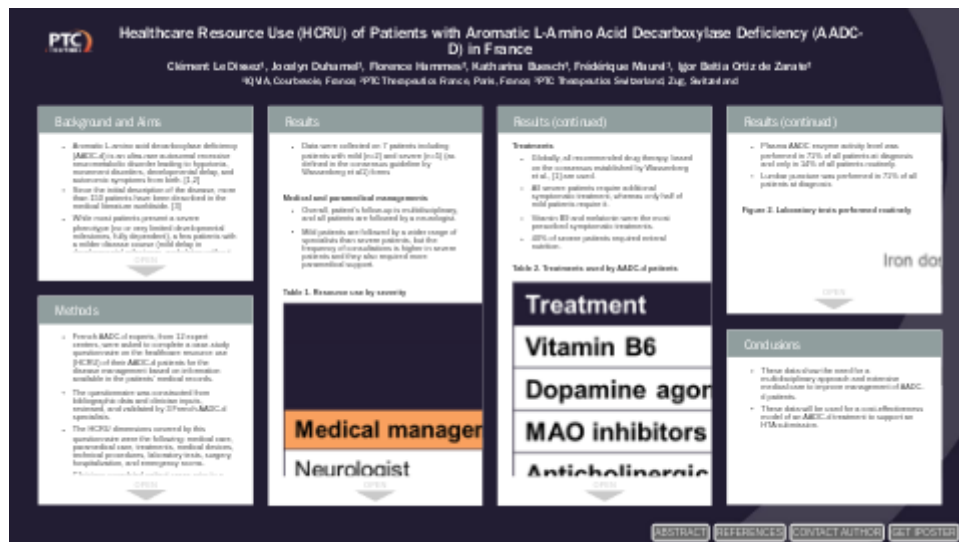


Healthcare Resource Use (HCRU) of Patients with Aromatic L-Amino Acid Decarboxylase Deficiency (AADC-D) in France



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PRESENTED AT:



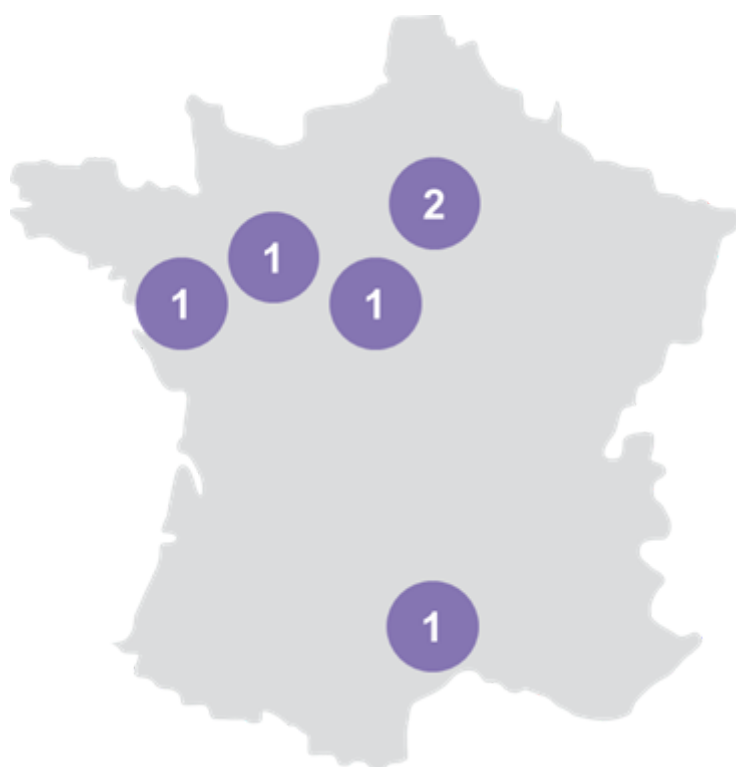
BACKGROUND AND AIMS

- Aromatic L-amino acid decarboxylase deficiency (AADC-d) is an ultra-rare autosomal recessive neurometabolic disorder leading to hypotonia, movement disorders, developmental delay, and autonomic symptoms from birth. [1,2]
- Since the initial description of the disease, more than 150 patients have been described in the medical literature worldwide. [3]
- While most patients present a severe phenotype (no or very limited developmental milestones, fully dependent), a few patients with a milder disease course (mild delay in developmental milestones, ambulatory without assistance, mild intellectual disability) are known. [1]
- Given the lack of healthcare resource utilization (HCRU) information available, the aim of this study, part of a global burden of disease project, was to generate French data to feed a cost-effectiveness model to support an HTA submission.

METHODS

- French AADC-d experts, from 12 expert centers, were asked to complete a case-study questionnaire on the healthcare resource use (HCRU) of their AADC-d patients for the disease management based on information available in the patients' medical records.
- The questionnaire was constructed from bibliographic data and clinician inputs, reviewed, and validated by 3 French AADC-d specialists.
- The HCRU dimensions covered by this questionnaire were the following: medical care, paramedical care, treatments, medical devices, technical procedures, laboratory tests, surgery, hospitalization, and emergency rooms.
- Clinicians completed patient cases prior to a telephone interview, which allowed them to discuss each patient case and to clarify specific points covered by the questionnaire.

Figure 1. Location of KOLs interviewed



RESULTS

- Data were collected on 7 patients including patients with mild (n=2) and severe (n=5) (as defined in the consensus guideline by Wassenberg et al) forms

Medical and paramedical managements

- Overall, patient's follow-up is multidisciplinary, and all patients are followed by a neurologist.
- Mild patients are followed by a wider range of specialists than severe patients, but the frequency of consultations is higher in severe patients and they also required more paramedical support.

Table 1. Resource use by severity

	Severe patients		Mild patients	
	% of patients	Frequency	% of patients	Frequency
Medical management				
Neurologist	100%	3.2/year	100%	2.5/year
General practitioner	80%	3.5/year	100%	3.0/year
Pediatrician	60%	4.0/year	50%	1.0/year
Ophthalmologist	40%	1.5/year	50%	1.0/year
Orthopedic Surgeon	60%	1.0/year	0%	-
Gastroenterologist	40%	1.0/year	0%	-
Physical medicine and rehabilitation specialist	20%	1.5/year	50%	2.0/year
Otolaryngologist	0%	-	50%	1.0/year
Psychiatrist	0%	-	50%	52/year
Orthopediatrician and orthopedist	0%	-	50%	2.0/year
Paramedical management				
Physiotherapist	100%	87/year	0%	-
Orthophonist	40%	65/year	100%	52/year
Occupational therapist	40%	2/year	50%	52/year
Dietetician	40%	4/year	50%	NR
Nurses	40%	NR	0%	-
Neuropsychological assessment	20%	NR	50%	3/year
Psychomotrician	40%	52/year	0%	-
Educator	20%	52/year	0%	-

RESULTS (CONTINUED)

Treatments

- Globally, all recommended drug therapy, based on the consensus established by Wassenberg et al., [1] are used.
- All severe patients require additional symptomatic treatment, whereas only half of mild patients require it.
- Vitamin B9 and melatonin were the most prescribed symptomatic treatments.
- 40% of severe patients required enteral nutrition.

Table 2. Treatments used by AADC-d patients

Treatment	Severe	Mild
Vitamin B6	100%	50%
Dopamine agonists	100%	100%
MAO inhibitors	80%	100%
Anticholinergic agents	20%	0%
Other treatments	100%	50%
<i>L-Dopa</i>	<i>20%</i>	<i>50%</i>
<i>Vitamin B9</i>	<i>80%</i>	<i>0%</i>
<i>Melatonin</i>	<i>60%</i>	<i>50%</i>
<i>Benzodiazepines</i>	<i>40%</i>	<i>0%</i>
<i>Dietary supplements</i>	<i>20%</i>	<i>50%</i>
<i>Enteral nutrition</i>	<i>40%</i>	<i>0%</i>
<i>Laroxyl drops</i>	<i>20%</i>	<i>0%</i>
<i>Uvedose</i>	<i>20%</i>	<i>0%</i>
<i>Ritalin</i>	<i>20%</i>	<i>0%</i>
<i>Parenteral nutrition</i>	<i>20%</i>	<i>0%</i>

Medical devices and Gastrostomy

- Medical device use was higher in severe patients (e.g., 100% required posterior molded shell, 80% needed a wheelchair, 60% needed a custom molded mattress).
- Medical device was only required for 50% of mild patients (lower limb splints and nocturnal orthotics).
- Gastrostomy (a result of significant impaired feeding) was common in severe patients (60%) but was absent in mild patients.

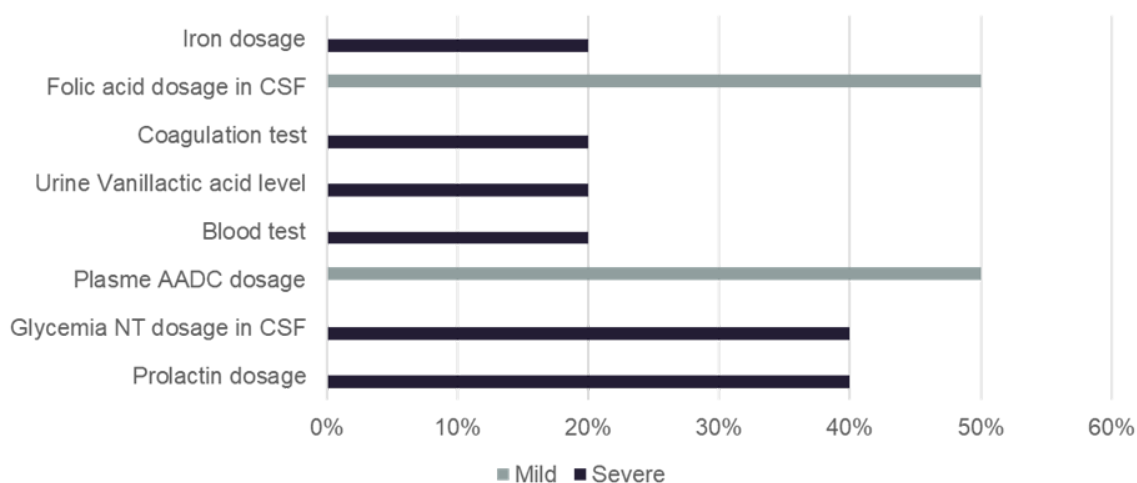
Laboratory tests and Medical procedures

- Most of the laboratory tests and medical procedures were performed at diagnosis but not on a routine basis.

RESULTS (CONTINUED)

- Plasma AADC enzyme activity level was performed in 71% of all patients at diagnosis and only in 14% of all patients routinely.
- Lumbar puncture was performed in 71% of all patients at diagnosis

Figure 2. Laboratory tests performed routinely



Hospitalizations

- Hospitalizations were more frequent in severe patients (80% had hospitalizations during previous 12 months with a mean number of 3 hospitalizations).
- Similarly, 80% of severe patients have had at least one emergency visit with a mean number of 2 visits.
- The main reasons for hospitalizations or emergency visits were breathing difficulties, lung infection and altered general condition.
- Only 50% of mild patients have had hospitalization (due to psychiatric conditions) but no emergency visit was required.

CONCLUSIONS

- These data show the need for a multidisciplinary approach and extensive medical care to improve management of AADC-d patients.
- These data will be used for a cost-effectiveness model of an AADC-d treatment to support an HTA submission.

ABSTRACT

HEALTHCARE RESOURCE USE (HCRU) OF PATIENT WITH AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY (AADC-D) IN FRANCE

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Objectives

AADC-d is an ultra-rare autosomal recessive neurometabolic disorder leading to hypotonia, movement disorders, developmental delay and autonomic symptoms from birth. Given the lack of healthcare resource utilization (HCRU) information, the aim of this study, part of a global burden of disease project, was to generate French data to feed a cost-effectiveness model to support HTA submission.

Methods

French AADC-d experts were asked to complete a case-study questionnaire on the HCRU of their AADC patients for the disease management based on information available in the patients' medical records. This questionnaire was constructed from bibliographic data and clinician inputs, reviewed and validated by 3 French AADC-d specialists. Clinicians completed patient cases prior to a telephone interview, which allowed them to discuss each patient case and to clarify specific points covered by the questionnaire.

Results

Data were collected on 7 patients including patients with mild (2) and severe (5) forms. Overall, patient follow-up is multidisciplinary. All patients are followed by a neurologist. Mild patients are followed by a larger panel of specialists than severe patients but the frequency of consultations is higher in severe patients and they also required more paramedical support. Globally, all recommended medications were used. Medical device use was higher in severe patients (e.g. 80% needed a wheelchair). Gastrostomy (a result of significant impaired feeding) was common in severe patients (60%) but was absent in mild patients. Most of the biochemical tests and medical procedures were performed at diagnosis but not on a routine basis. Finally, hospitalizations were more frequent in severe patients (80% had hospitalizations during previous 12 months with a mean number of 3 hospitalizations).

Conclusions

These data show the need of a multidisciplinary approach and extensive medical care to manage AADC-d patients. These data will be used for a cost-effectiveness model of an AADC-d treatment to support an HTA submission.

REFERENCES

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