

# SUMMARY

- We report the clinical, radiological and biochemical findings in 18 patients (16 families). All patients were examined neurologically. Nuclear Magnetic Resonance Spectroscopy was performed in three patients, Cerebral Magnetic Resonance in 16 cases, and Serum levels for very long chain fatty acids (VLCFA) were determined in plasma samples of all the individuals and in 30 family relatives. 17 patients presented neurological degenerative disease signs, two of them had adrenal dysfunction and one was asymptomatic (considered a possible bone marrow donor for affected brother).
- The C26:0 concentrations ranged from 0.740 to 1.320  $\mu\text{g/ml}$  (VR:  $0.22 \pm 0.08$ ). The C24/C22 ratio was from 1.751 to 2.198 (VR:  $0.84 \pm 0.08$ ) and the C26/C22 was from 0.072 to 0.167 (VR:  $0.01 \pm 0.01$ ). In summary we have found abnormal VLCFA values in 11 cases indicating X-ADL, seven obligate heterozygote and five other asymptomatic family relatives with values compatible with the heterozygote state.

# OBJECTIVE

- The main goal of the present work, was to identify and to verify biochemically, by determination of VLCFA levels, the cases of X-ALD in the group of patients sent for degenerative diseases study to the Instituto de Errores Innatos del Metabolismo - Pontificia Universidad Javeriana

# METHODS

- All patients were examined neurologically. Nuclear Magnetic Resonance Spectroscopy was performed in three patients, Cerebral Magnetic Resonance in 16 cases, and serum levels for very long chain fatty acids by gas chromatography-mass spectrometry were determined in all the individuals as well as in 30 family members.

# CONCLUSIONS

- After a year of follow-up most patients presented gait disturbances and lack of coordination. One patient was diagnosed in the neonatal stage and one in adulthood whom, at the time of diagnosis, had adrenal insufficiency. Interestingly we found a patient's mother with normal VLCFA levels.
- In this study we reported for the first time several cases of X-ALD in the Colombian population. Although at the moment none of the patients has had access to palliative treatment, the possibility to detect the disease without symptoms allows for treatment when it becomes available.

# ACKNOWLEDGMENTS

- The authors are grateful to Dr Moser's team, specially to Dr Steven Steinberg in the Kennedy Klieger Institute for all the technical assistance.
- They also wish to thank the members of the Instituto de Errores Innatos del Metabolismo for their kind contributions to this work.



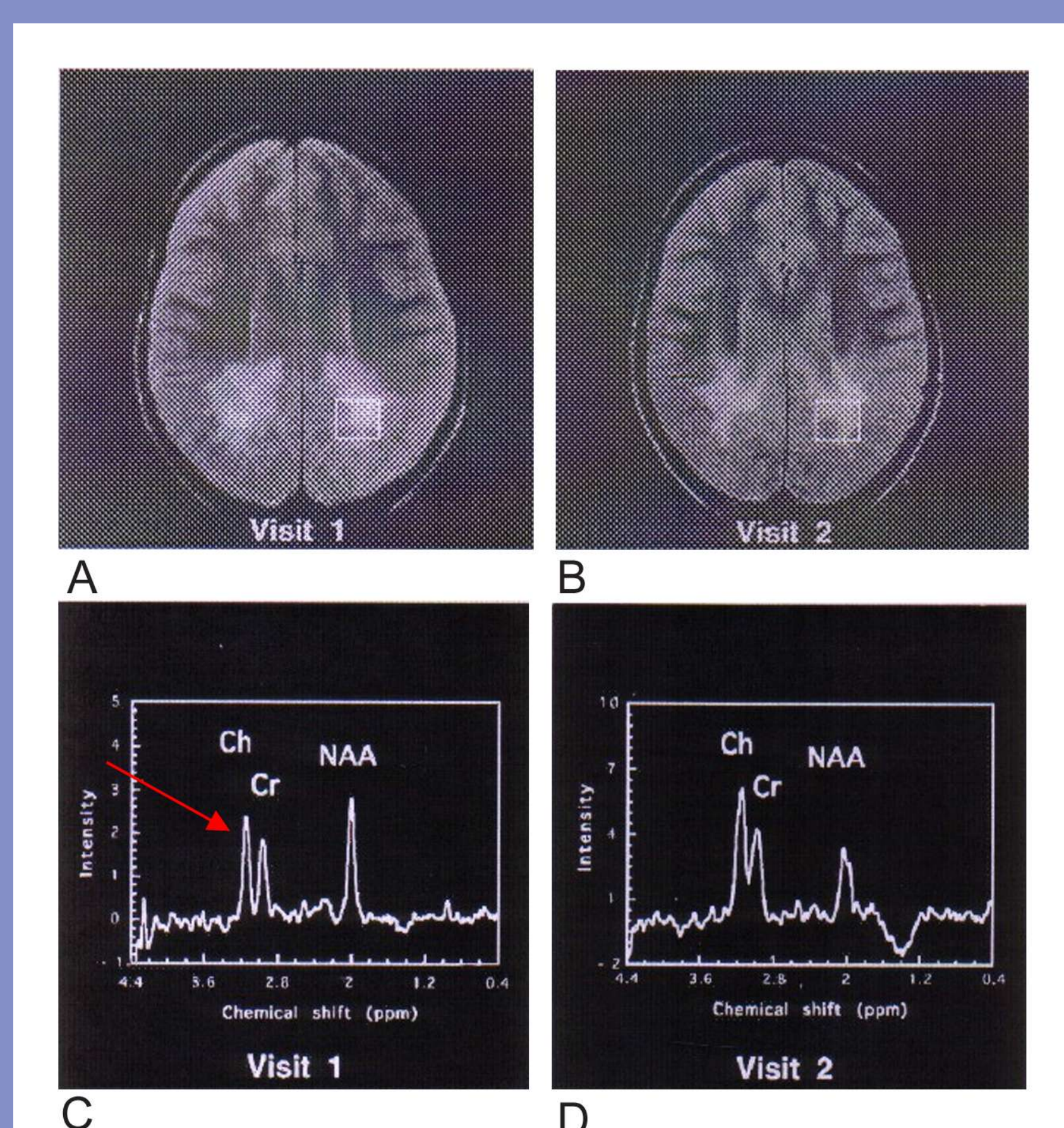
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# X-LINKED ADRENOLEUKODYSTROPHY THE COLOMBIAN EXPERIENCE

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# RESULTS



MRS was performed in three cases. The choline peak was dramatically elevated.

Table 1. VLCFA levels in studied patients

	AGE (years)	C26:0 (vr. $0.22 \pm 0.08$ )	RATE C24/C22 (vr. $0.84 \pm 0.08$ )	RATE C26/C22 (vr. $0.01 \pm 0.01$ )
CASE 1	24	1.145	1.923	0.107
CASE 2	9	0.740	1.971	0.072
CASE 3	5	1.370	1.735	0.107
CASE 4	12	1.320	2.198	0.121
CASE 5	10	0.971	1.751	0.089
CASE 6	6	0.970	1.851	0.167
CASE 7	9	0.970	1.851	0.167
CASE 8	26	1.010	1.774	0.060
CASE 9	2	1.310	1.7	0.080
CASE 10	12	1.410	1.999	0.121
CASE 11	9	0.740	1.971	0.072
CASE 12	12	1.320	2.198	0.121
CASE 13	15	1.059	1.968	0.099
CASE 14	11	1.231	1.878	0.114
CASE 15	5	1.643	1.955	0.175
CASE 16	7	1.626	2.014	0.205

Table 2. Main clinical manifestations in affected subjects

Average age of diagnosis (years)	14.6
Coma	5/16
Seizures	11/16
Muscular weakness	6/16
Deafness	10/16
Visual defects	12/16
Hyperactivity	4/16
Urinary disorders	5/16
Spatial disorientation	10/16
Behaviour disturbances	6/16
Gait disturbances	12/16
Adrenal compromise	3/16
Asymptomatic	1/16

