

Neues
aus Forschung und Wissenschaft
zur Galaktosämie

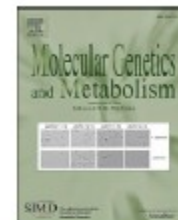


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How strict is galactose restriction in adults with galactosaemia? International practice



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Table 1
Dietary treatment policy and diet relaxation practices for adults with galactosaemia reported by centres.

Country represented by participating centres	No. of centres from each country	Total number of patients	Dietary treatment policy – restricted foods				Allow relaxed diet	Types of foods permitted in relaxed diet
			Lactose	Galactose from fruit and vegetables	Galactose from galactosides	Galactose restricted from offal		
UK	11	109	11/11	0/11	0/11	0/11	4/11	Lactose containing ingredients in manufactured foods
Germany	6	65	6/6	0/6	0/6	4/6	2/6	Lactose containing ingredients in manufactured foods
Netherlands	5	51	5/5	0/5	0/5	0/5	3/5	Lactose containing ingredients in manufactured foods
Belgium	4	21	4/4	1/4	1/4	1/4	3/4	3/4 centres lactose containing ingredients in manufactured foods
Australia	2	54	2/2	0/2	1/2	1/2	0/2	None
France	2	21	2/2	0/2	0/2	0/2	2/2	Diet relaxed to a lactose-free diet only
Italy	2	19	2/2	2/2	2/2	2/2	0/2	None
Spain	2	16	2/2	1/2	2/2	2/2	0/2	None
Sweden	2	11	2/2	0/2	0/2	1/2	0/2	None
New Zealand	1	44	1/1	0/1	0/1	0/1	1/1	Lactose containing ingredients in manufactured foods
Norway	1	4	1/1	0/1	0/1	0/1	0/1	None
Switzerland	1	3	1/1	0/1	0/1	1/1	0/1	None
TOTAL	39	418	39/39 (100%)	4/39 (10%)	6/39 (15%)	12/39 (31%)	15/39 (38%)	

Definitions: *lactose restriction*: avoidances of animal milk and products contains animal milk e.g. yoghurt, ice cream, lactose e.g. tomatoes, kiwi fruits, banana; *avoidance of galactosides* e.g. peas, beans, lentils, cocoa; *restriction of galactose storage or*

† high galactose containing fruit/vegetables

Effects of temporary low-dose galactose supplements in children aged 5–12 y with classical galactosemia: a pilot study

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BACKGROUND: Classical galactosemia is caused by severe galactose-1-phosphate uridylyltransferase deficiency. Despite life-long galactose-restriction, many patients experience long-term complications. Intoxication by galactose and its

impairment, speech and language abnormalities, tremor, ataxia, osteopenia, and over 80% of females have premature ovarian insufficiency (2–4). The pathophysiology is complex and comprises toxic build-up of galactose and its metabolites

Belastungsgruppe

13 Patienten

5 – 11 Jahre

Q188R-Homozygot

Kontrollgruppe

13 Patienten

5 – 11 Jahre

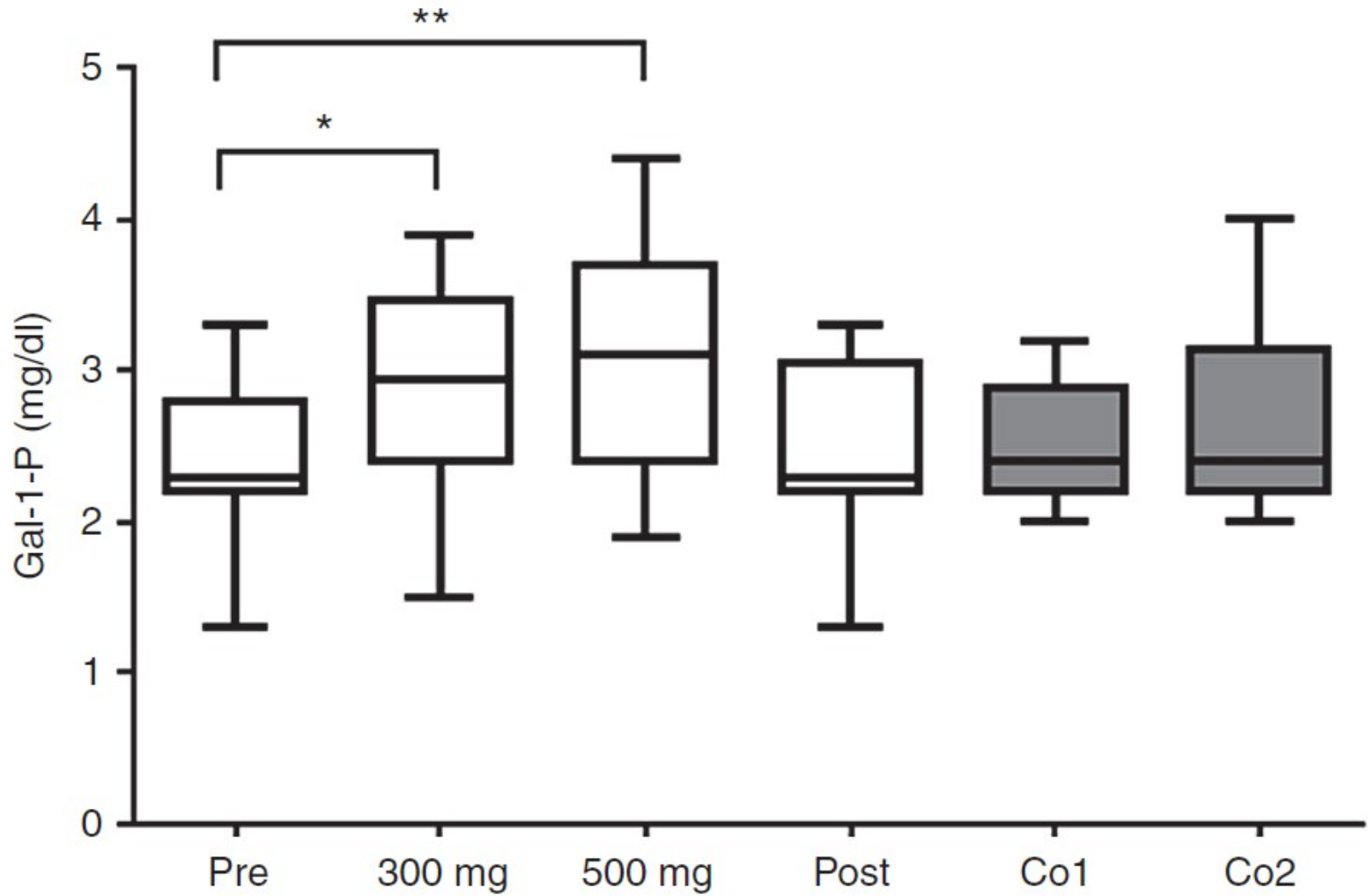
Q188R-Homozygot

Galaktosebelastung durch Gabe von Milch

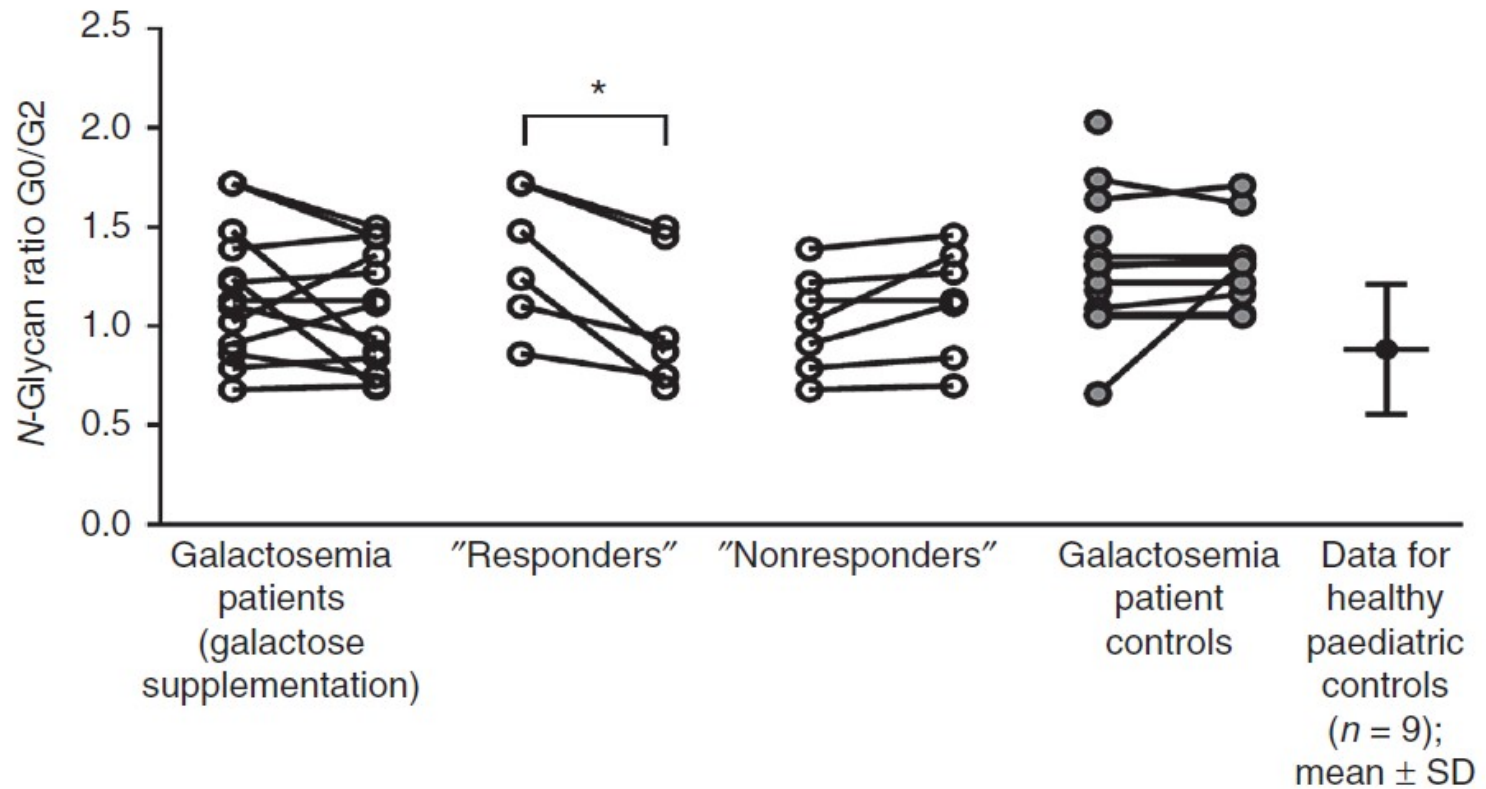
Galaktosedosis 0 – 300 – 500 – 0 mg/Tag

jeweils 2 Wochen pro Periode

Stoffwechselfparameter Gal-1-P



Galatosylierung IgG im Plasma



Motor and Speech Disorders in Classic Galactosemia

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Lawrence D. Shriberg

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Abstract *Purpose* To test the hypothesis that children with classic galactosemia and speech disorders are at risk for co-occurring strength and coordination disorders.

Method This is a case–control study of 32 children (66% male) with galactosemia and neurologic speech disorders and 120 controls (50% male) ages 4–16 years. Speech was

($n = 7$) and ataxic dysarthria ($n = 1$), had poorer balance and manual dexterity, but not weaker hand or tongue strength, compared to the children with fewer speech errors. The number of days on milk during the neonatal period was associated with more speech errors in males but not in females.

Kollektive

32 Patienten, 4 – 16 Jahre

130 Kontrollen, 4 – 16 Jahre

Testbatterien

Sprachtests

Zungen- und Handstärkenmessung

Bewegungs- und Koordinationstests

Conclusion Children with galactosemia have a high prevalence of co-occurring speech, coordination, and strength disorders, which may be evidence of a common underlying etiology, likely associated with diffuse cerebellar damage, rather than distinct disorders.

Schlussfolgerung: Kinder mit Galaktosämie haben häufig gleichzeitig auftretende Problem mit Sprachbildung, Koordination, und Muskelstärke. Dies dürfte auf eine gemeinsame Ursache hinweisen, die wahrscheinlich eher mit einer diffusen cerebralen Schädigung assoziiert ist als mit spezifischen Schädigungen.

White matter microstructure pathology in classic galactosemia revealed by neurite orientation dispersion and density imaging

Inge Timmers • Hui Zhang • Matteo Bastiani •
Bernadette M. Jansma • Alard Roebroek •
M. Estela Rubio-Gozalbo

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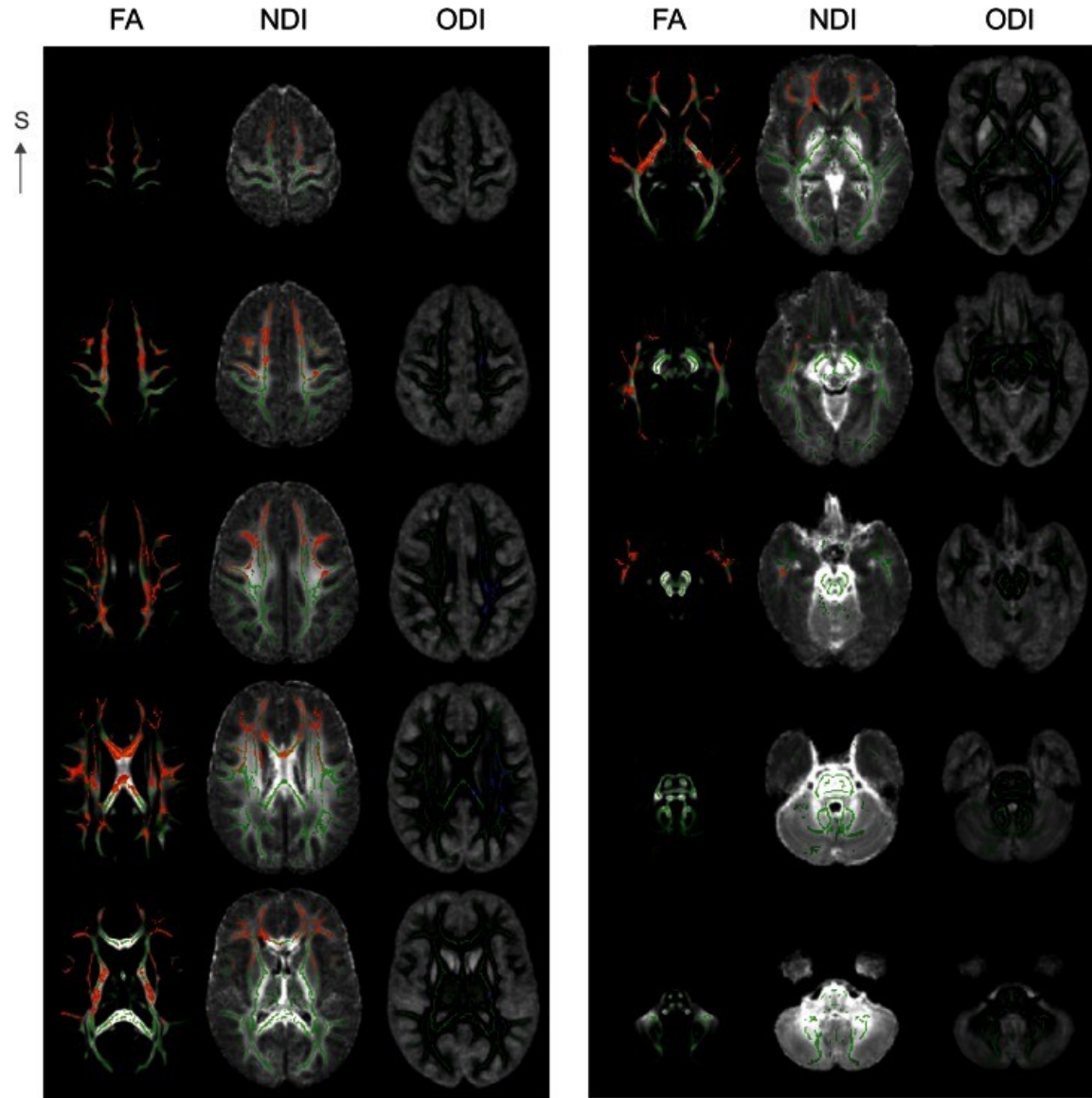
Abstract White matter abnormalities have been observed in patients with classic galactosemia, an inborn error of galactose metabolism. However, magnetic resonance imaging (MRI) data collected in the past were generally qualitative in nature. Our objective was to investigate white matter microstructure

are in agreement with the cognitive profile observed in galactosemia, showing higher order cognitive impairments, and language and motor impairments, respectively. Less favourable white matter properties correlated positively with age and age at onset of diet, and negatively with behavioural outcome (e.g.

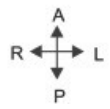
MRI bei

Patienten (16 - 21 Jahre, n = 8)

Kontrollen (15 - 20 Jahre, n = 8)



FA
 Anisotropie
NDI
 Neuritendichte
ODI
 Orientationsdispersion



patients > controls
controls > patients



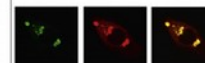
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Research Report

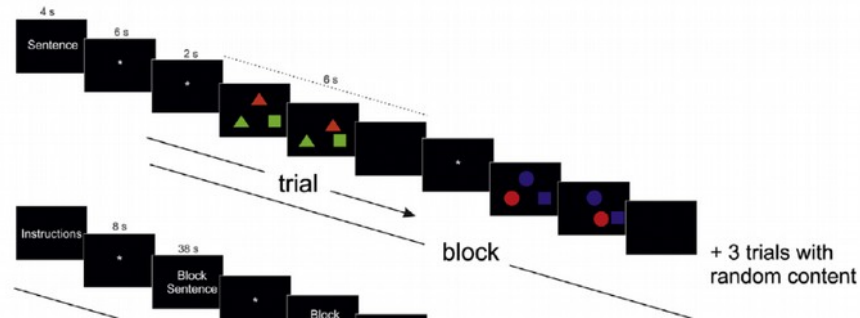
Affected functional networks associated with sentence production in classic galactosemia



Inge Timmers^{a,b}, Job van den Hurk^{b,c,d}, Paul AM Hofman^e,
Luc JI Zimmermann^a, Kâmil Uludağ^{b,c}, Bernadette M Jansma^{b,c},
M Estela Rubio-Gozalbo^{a,f,*}

fMRI

Satzbildungstest bei Patienten und gesunden Kontrollen (n = 13)



Group differences in the contrast language production versus baseline

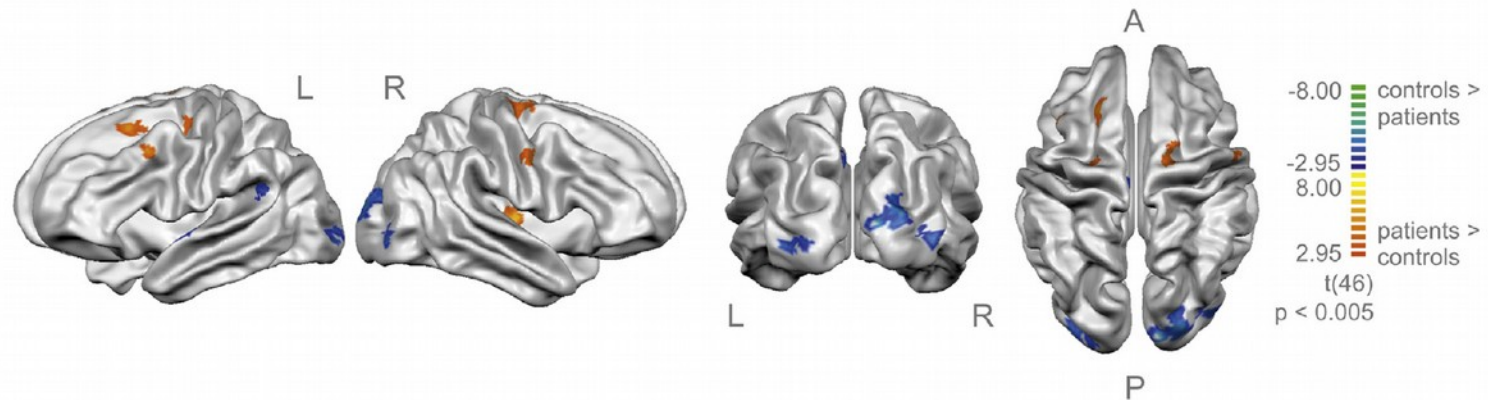


Fig. 3 – Group differences across the cortical surface in the contrast language production versus baseline [$'S'+W' > \text{baseline}$]. In red, areas are shown in which patients showed higher BOLD signal change compared to controls; in blue regions, the patients showed lower signal change compared to controls. (For interpretation of the references to color in this figure legend, the reader is referred to the web version of this article.)

Arginine Functionally Improves Clinically Relevant Human Galactose-1-Phosphate Uridylyltransferase (GALT) Variants Expressed in a Prokaryotic Model

Ana L. Coelho • Matilde Trabuco • Maria João Silva •
Isabel Tavares de Almeida • Paula Leandro •
Isabel Rivera • João B. Vicente

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Abstract Classic galactosemia is a rare genetic disease of the galactose metabolism, resulting from deficient activity of galactose-1-phosphate uridylyltransferase (GALT). The current standard of care is lifelong dietary restriction of

Furthermore, it revealed that arginine presents a mutation-specific beneficial effect, particularly on the prevalent p.Q188R and p.K285N variants, which led us to hypothesize that it might constitute a promising therapeutic agent in