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CASE REPORT

Severe disseminated mycobacterial infection in a boy with a novel mutation leading to IFN-γR2 deficiency

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KEYWORDS
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IFN-γR2;
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Mutation;
Immunodeficiency;
IFNGR2;
Primary immunodeficiency

Summary Mendelian susceptibility to mycobacterial diseases (MSMD) is a rare syndrome characterized by predisposition to severe, sometimes lethal, disease caused by otherwise poorly virulent mycobacteria. We report here a boy with a recurrent mycobacterial infection from the age of five months. Immunological analyses revealed an inability to respond to IFN-γ, subsequent genetic analyses revealed a novel homozygous mutation, r.679G > A in the *IFNGR2* gene, resulting in a G227R substitution, that caused IFN-γR2 deficiency. This is only the 8th mutation in IFN-γR2 known so far. The boy eventually died of hepatic coma due to liver failure at the age of five.

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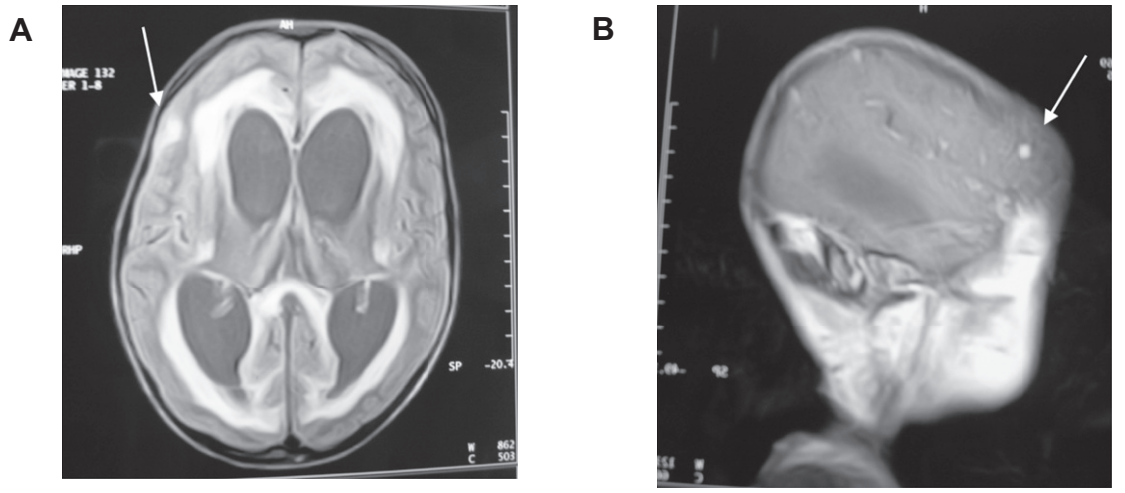
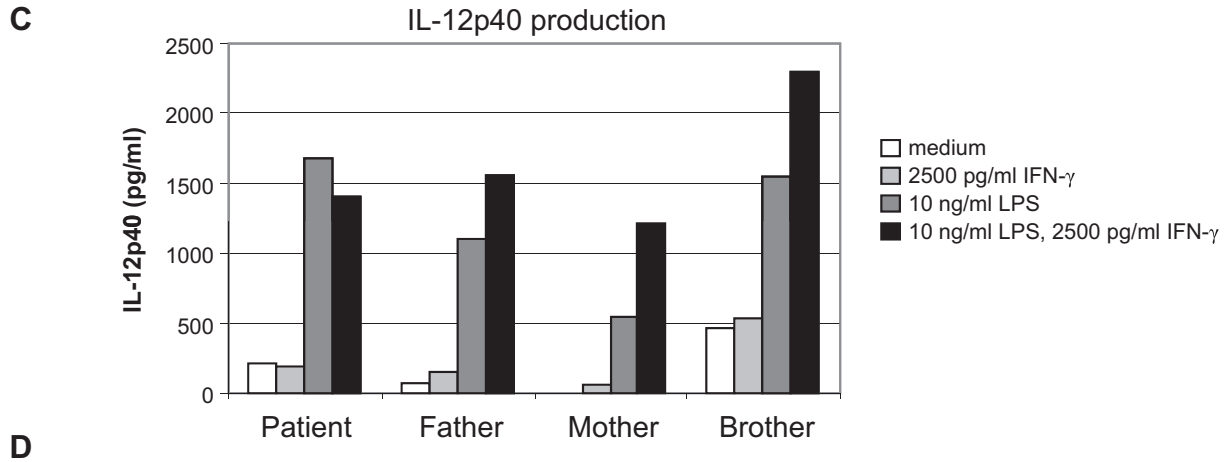
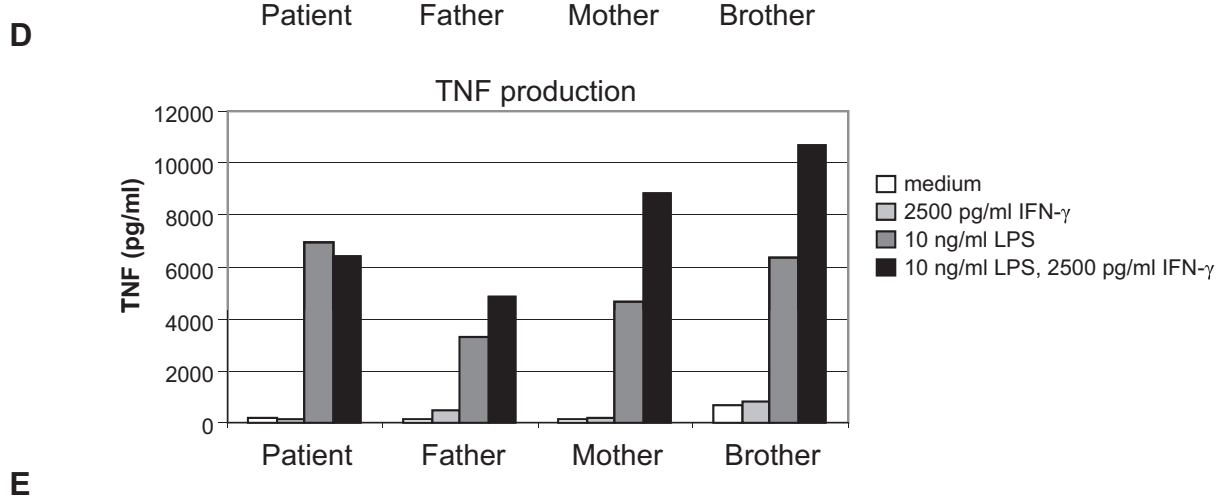
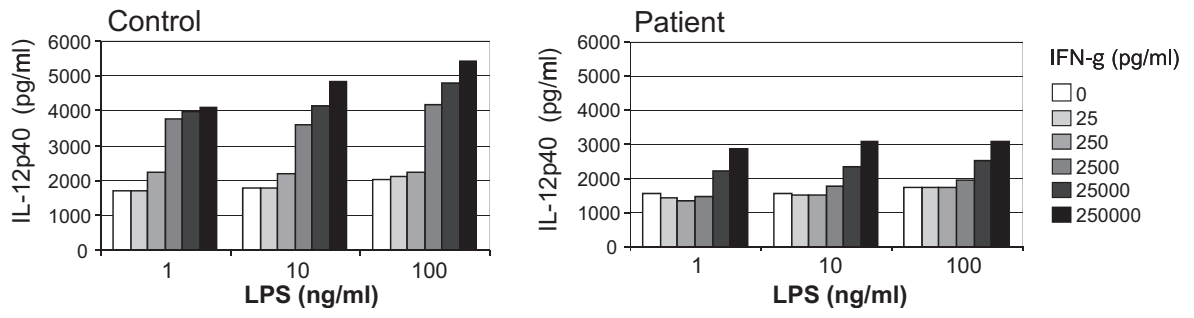

A
B


C
IL-12p40 production
IL-12p40 (pg/ml)
2500
2000
1500
1000
500
0
Patient
Father
Mother
Brother
medium
2500 pg/ml IFN-γ
10 ng/ml LPS
10 ng/ml LPS, 2500 pg/ml IFN-γ


D
TNF production
TNF (pg/ml)
12000
10000
8000
6000
4000
2000
0
Patient
Father
Mother
Brother
medium
2500 pg/ml IFN-γ
10 ng/ml LPS
10 ng/ml LPS, 2500 pg/ml IFN-γ


E
Control
Patient
IL-12p40 (pg/ml)
6000
5000
4000
3000
2000
1000
0
1
10
100
LPS (ng/ml)
IFN-g (pg/ml)
0
25
250
2500
25000
250000