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12.3 Long-term follow up of patients with CINCA syndrome: efficacy and tolerability of Anakinra treatment

G Paloni*¹, M Gattorno², M Alessio³, D Rigante⁴, M Cattalini⁵, F Zulian⁶ and L Lepore¹

Address: ¹Institute of Child Health IRCCS Burlo Garofolo, University of Trieste, Italy, ²II Division of Pediatrics, 'G. Gaslini' Institute for Children, Genoa, Italy, ³Department of Pediatrics, Federico II Hospital, Naples, ⁴Department of Pediatric Sciences, Catholic University of Sacred Heart, Rome, Italy, ⁵Department of Rheumatology and Clinical Immunology, Brescia, Italy and ⁶Pediatric Rheumatology Unit, Department of Pediatrics, Padova, Italy

* Corresponding author

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We report the long-term follow up of 15 Italian patients affected by Chronic Infantile Neurological Cutaneous and Articular (CINCA) syndrome (7 males and 8 females, mean age 13, 5 years).

We report the updating of the survey following the introduction of the IL-1 receptor antagonist therapy (Anakinra).

11 patients were treated with subcutaneous injections, at a daily dosage of 1 mg/Kg in children and up to 100 mg in adults, 2 refused the therapy, and the others did not require the treatment due to a mild presentation of the disease so far.

In treated patients fever, rash, articular involvement, conjunctivitis, uveitis disappeared or improved. Laboratory data normalized in most of the cases within the first month. The neurologic symptoms ameliorated, papilledema disappeared in four out of eleven treated patients while dysmorphisms (typical facies) and bone alterations constantly persist. No further Cinca related signs and symptoms appeared after Anakinra was introduced.

The medication showed very good tolerability: we observed local erythema at the site of injection in 2 out of 11 subjects, and oral aphthosis in another.

The untreated subjects kept presenting all their symptoms of CINCA syndrome and the disease went on with its poor manifestation. We found no differences in the response to Anakinra therapy between *CIAS1*-mutated and not mutated patients.

In summary, our study demonstrates that a long lasting treatment with Anakinra appears to be safe and highly effective in patients affected by CINCA syndrome.