Appendix e-1

**Transcriptome assay and data analysis.** For each patient (5 AT, 3 females and 2 males, average age of 15 years), healthy subjects ( childhood blood samples, unknown gender) and the case report, 3 ml of whole blood was collected by Tempus Blood RNA tubes (Applied Biosystems). The blood drawn was achieved only in patient’s healthy state. Total RNA was gotten by Tempus spin RNA isolation kit (Applied Biosystems) and it was labelled using the MessageAmp™ II-Biotin Enhanced aRNA Amplification Kit (Ambion, Austin, TX, USA) according to the manufacturer’s instructions.

Gene expression was assessed on the CodeLink Human Whole Genome Bioarray, and the post processing was performed by GeneSifter (www.genesifter.net; Geospiza Inc., Seattle, WA, USA). Normalized data were analysed by MeV (7) software package, by using a permutation based t-test (p≤0.01) between A-T and WT samples groups, skipping the expression profile of the case report patient, that was subsequently re-introduced as unknown sample in the Hierarchical Clustering (HLC) computation (Pearson correlation metric) in order to categorize it.

The HLC process allows the estimation of sample similarity by gene expression extent of statistically differentially expressed probes (1326). The graph reports the similarity dendrogram of samples, and the coloured drawing of expression level of each probe.

In order to establish the probe expression similarity of the case report to WT or A-T group, each average probe expression of each group was calculated as reference. The smallest Euclidean distance expression value, between the case report data and each reference, was used to categorize the atypical case (applying a 1.2 fold change cut-off filter).

The relationships among differentially expressed genes were explored by performing a network-based analysis using the Reactome Functional Interaction (FI) Network plugin for Cytoscape.