Raising Awareness of the Rare Disease Sanfilippo Syndrome C Using The Open Drug Discovery Teams (ODDT) Mobile App

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What is Sanfilippo disease type C?
Mucopolysaccharidosis Type IIIC (MPS IIIC or Sanfilippo disease type C) is one such LSD that is caused by deficiency of enzyme heparan sulfate acetyl CoA: a-glucosaminide N-acetyltransferase, (HGSNAT) responsible for degradation of heparan sulfate, a repeating carbohydrate generally found attached to proteoglycans. The clinical phenotype includes onset in infancy or early childhood, progressive and severe neurological deterioration causing hyperactivity, sleep disorders and loss of speech accompanied by behavioral abnormalities, neuropsychiatric problems, mental retardation, hearing loss, and visceral manifestations, such as mild hepatomegaly, joint stiffness, vertebral bodies and hypertrichosis.

There is no treatment but there are research efforts to find one funded primarily by multiple disease foundations. The number of patients and research community engaged are both small. The amount of research funding available is limited so any efforts to raise awareness of this disease are key.

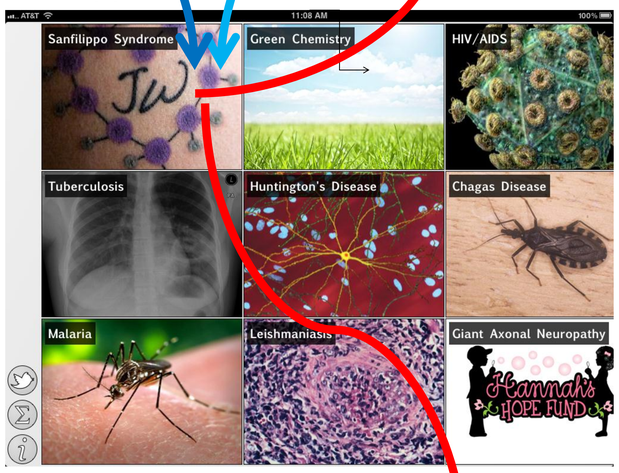
The Open Drug Discovery Teams (ODDT) project uses a free mobile app as user entry point **http://tinyurl.com/6l9qy4f**. The app has a magazine-like interface, and server-side infrastructure for hosting chemistry-related data as well as value added services. The project is open to participation from anyone and provides the ability for users to make annotations and assertions, thereby contributing to the collective value of the data to the engaged community. The infrastructure for the app is currently based upon the Twitter API and uses Google Alerts RSS feeds as a useful proof of concept for a real time source of publicly generated content.

We now highlight how ODDT can be used to raise awareness of Sanfilippo syndrome and engage this disease community by following #sanfilipposyndrome using Twitter and Google Alerts.

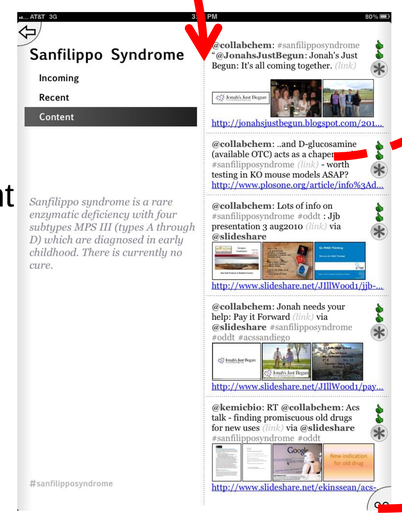
@jonahsjustbegun and @collabchem tweets with #sanfilipposyndrome

Content from web Google Alerts

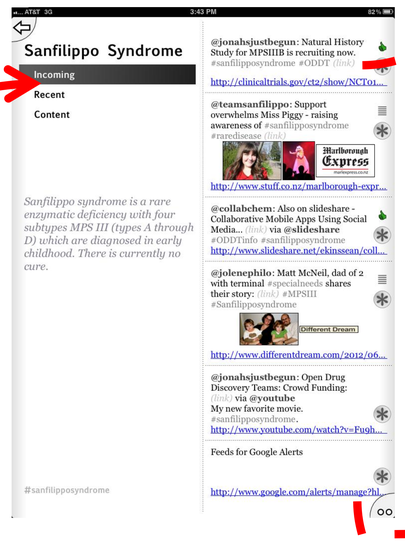
The App



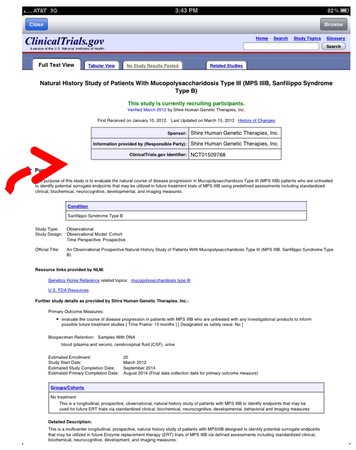
Items that are endorsed are saved to content tab



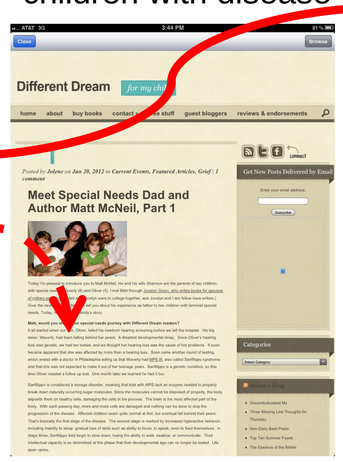
Content collected ready for viewing



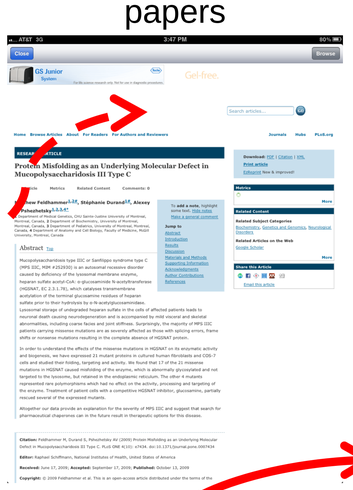
Raise awareness of clinical studies

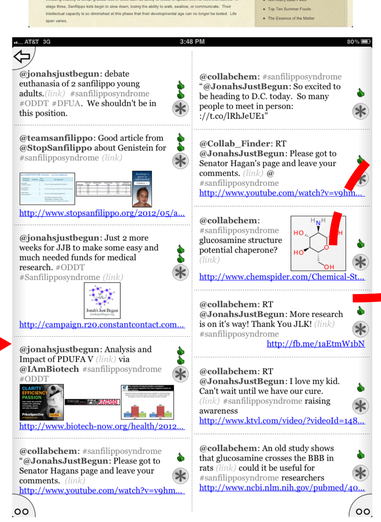


Raise awareness of children with disease



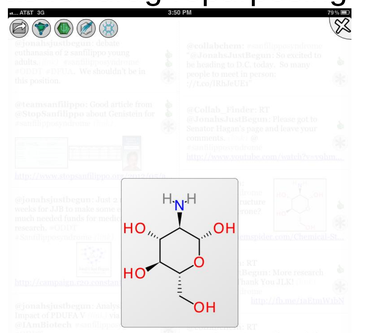
Highlight and link to important published papers



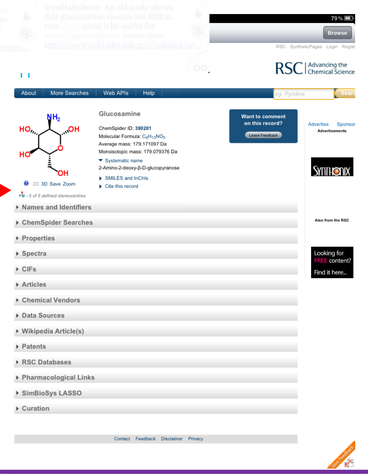


Content collected ready for viewing

Share molecules and ideas for drug repurposing



Links to other databases



Information on rare disease advocacy

Highlight availability and cost of treatments for rare diseases